

Christopher E Shaw

List of Publications by Year in descending order

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Version: 2025-02-01

203
papers

34,647
citations

4704

80
h-index

2486

185
g-index

223
all docs

223
docs citations

223
times ranked

31348
citing authors

#	ARTICLE	IF	CITATIONS
1	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. <i>Npj Genomic Medicine</i> , 2022, 7, .	4.5	31
2	Disruption of ERâ€mitochondria tethering and signalling in <i>C9orf72</i>â€associated amyotrophic lateral sclerosis and frontotemporal dementia. <i>Aging Cell</i> , 2022, 21, .	7.0	42
3	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. <i>Circulation Research</i> , 2022, 130, 166-180.	12.8	25
4	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. <i>Science Translational Medicine</i> , 2022, 14, .	13.1	55
5	C9orf72 poly GA RAN-translated protein plays a key role in amyotrophic lateral sclerosis via aggregation and toxicity. <i>Human Molecular Genetics</i> , 2021, 30, 318-320.	3.1	4
6	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , 2021, 109, 448-460.e4.	12.8	65
7	Generation of six induced pluripotent stem cell lines from patients with amyotrophic lateral sclerosis with associated genetic mutations in either FUS or ANXA11. <i>Stem Cell Research</i> , 2021, 52, 102246.	0.6	3
8	Disease Mechanisms and Therapeutic Approaches in C9orf72 ALS-FTD. <i>Biomedicines</i> , 2021, 9, 601.	3.6	9
9	ALS-linked FUS mutants affect the localization of U7 snRNP and replication-dependent histone gene expression in human cells. <i>Scientific Reports</i> , 2021, 11, .	3.7	5
10	Regulation of Synapse Weakening through Interactions of the Microtubule Associated Protein Tau with PACSIN1. <i>Journal of Neuroscience</i> , 2021, 41, 7162-7170.	3.7	12
11	Cytoplasmic TDP-43 is involved in cell fate during stress recovery. <i>Human Molecular Genetics</i> , 2021, 31, 166-175.	3.1	15
12	Demystifying the spontaneous phenomena of motor hyperexcitability. <i>Clinical Neurophysiology</i> , 2021, 132, 1830-1844.	1.0	16
13	Elucidating the Role of Cerebellar Synaptic Dysfunction in C9orf72-ALS/FTD â€” a Systematic Review and Meta-Analysis. <i>Cerebellum</i> , 2021, 21, 681-714.	2.1	6
14	A recessive S174X mutation in Optineurin causes amyotrophic lateral sclerosis through a loss of function via allele-specific nonsense-mediated decay. <i>Neurobiology of Aging</i> , 2021, 106, 1-6.	3.4	4
15	<i>SCFD1</i> expression quantitative trait loci in amyotrophic lateral sclerosis are differentially expressed. <i>Brain Communications</i> , 2021, 3, .	3.8	13
16	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	16.3	246
17	Mutant <i>C9orf72</i> human iPSCâ€derived astrocytes cause nonâ€cell autonomous motor neuron pathophysiology. <i>Glia</i> , 2020, 68, 1046-1064.	5.2	86
18	The use of biotelemetry to explore disease progression markers in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 563-573.	2.9	16

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19	ALS/FTD mutations in UBQLN2 impede autophagy by reducing autophagosome acidification through loss of function. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 15230-15241.	7.7	60
20	CYLD is a causative gene for frontotemporal dementia “ amyotrophic lateral sclerosis. <i>Brain</i> , 2020, 143, 783-799.	8.9	59
21	<i>ATXN1</i> repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. <i>Brain Communications</i> , 2020, 2, .	3.8	36
22	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. <i>NAR Genomics and Bioinformatics</i> , 2020, 2, .	2.3	13
23	C9orf72 intermediate expansions of 24“30 repeats are associated with ALS. <i>Acta Neuropathologica Communications</i> , 2019, 7, .	5.1	72
24	RRM adjacent TARDBP mutations disrupt RNA binding and enhance TDP-43 proteinopathy. <i>Brain</i> , 2019, 142, 3753-3770.	8.9	66
25	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. <i>Frontiers in Neuroscience</i> , 2019, 13, .	3.0	13
26	Relative preservation of triceps over biceps strength in upper limb-onset ALS: the “split elbow“™. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 730-733.	2.0	36
27	Predicting the future of ALS: the impact of demographic change and potential new treatments on the prevalence of ALS in the United Kingdom, 2020“2116. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 264-274.	2.9	22
28	Telomere length is greater in ALS than in controls: a whole genome sequencing study. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 229-234.	2.9	20
29	Nuclear RNA foci from<i>C9ORF72</i> expansion mutation form paraspeckle-like bodies. <i>Journal of Cell Science</i> , 2019, , .	3.2	31
30	Genetic meta-analysis of diagnosed Alzheimer“™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019, 51, 414-430.	16.3	1,802
31	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. <i>Nature Neuroscience</i> , 2019, 22, 1966-1974.	12.4	96
32	Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. <i>Neurobiology of Aging</i> , 2019, 74, 234.e9-234.e15.	3.4	26
33	Striking phenotypic variation in a family with the P506S UBQLN2 mutation including amyotrophic lateral sclerosis, spastic paraplegia, and frontotemporal dementia. <i>Neurobiology of Aging</i> , 2019, 73, 229.e5-229.e9.	3.4	13
34	Younger age of onset in familial amyotrophic lateral sclerosis is a result of pathogenic gene variants, rather than ascertainment bias. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 268-271.	2.0	38
35	Objectively Monitoring Amyotrophic Lateral Sclerosis Patient Symptoms During Clinical Trials With Sensors: Observational Study. <i>JMIR MHealth and UHealth</i> , 2019, 7, e13433.	5.5	35
36	C9ORF72 repeat expansion causes vulnerability of motor neurons to Ca2+-permeable AMPA receptor-mediated excitotoxicity. <i>Nature Communications</i> , 2018, 9, .	14.1	142

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37	Mitochondrial abnormalities and disruption of the neuromuscular junction precede the clinical phenotype and motor neuron loss in hFUSWT transgenic mice. <i>Human Molecular Genetics</i> , 2018, 27, 463-474.	3.1	71
38	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. <i>Lancet Neurology</i> , The, 2018, 17, 423-433.	19.1	372
39	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1267-1288.	12.8	482
40	A feedback loop between dipeptide-repeat protein, TDP-43 and karyopherin- β mediates C9orf72-related neurodegeneration. <i>Brain</i> , 2018, 141, 2908-2924.	8.9	67
41	ALS/FTD-Linked Mutation in FUS Suppresses Intra-axonal Protein Synthesis and Drives Disease Without Nuclear Loss-of-Function of FUS. <i>Neuron</i> , 2018, 100, 816-830.e7.	12.8	162
42	ALS-specific cognitive and behavior changes associated with advancing disease stage in ALS. <i>Neurology</i> , 2018, 91, .	1.3	175
43	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. <i>Neurobiology of Aging</i> , 2018, 71, 266.e1-266.e10.	3.4	61
44	TDP-43 causes neurotoxicity and cytoskeletal dysfunction in primary cortical neurons. <i>PLoS ONE</i> , 2018, 13, e0196528.	2.5	23
45	Amyotrophic Lateral Sclerosis and Other TDP-43 Proteinopathies. , 2018, , 99-115.		1
46	ATXN2 trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9.	3.4	96
47	C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 281.1-281.	2.0	34
48	Non-nuclear Pool of Splicing Factor SFPQ Regulates Axonal Transcripts Required for Normal Motor Development. <i>Neuron</i> , 2017, 94, 322-336.e5.	12.8	53
49	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. <i>Brain</i> , 2017, 140, 1611-1618.	8.9	66
50	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, .	13.1	127
51	Amyotrophic lateral sclerosis-like superoxide dismutase 1 proteinopathy is associated with neuronal loss in Parkinson's disease brain. <i>Acta Neuropathologica</i> , 2017, 134, 113-127.	7.9	80
52	C9orf72 poly GA RAN-translated protein plays a key role in amyotrophic lateral sclerosis via aggregation and toxicity. <i>Human Molecular Genetics</i> , 2017, 26, 4765-4777.	3.1	66
53	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. <i>Neurology</i> , 2017, 89, 1915-1922.	1.3	81
54	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. <i>Nature Genetics</i> , 2017, 49, 1373-1384.	16.3	683

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55	The benefit of evolving multidisciplinary care in ALS: a diagnostic cohort survival comparison. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 569-575.	2.9	38
56	C9ORF72 and UBQLN2 mutations are causes of amyotrophic lateral sclerosis in New Zealand: a genetic and pathologic study using banked human brain tissue. <i>Neurobiology of Aging</i> , 2017, 49, 214.e1-214.e5.	3.4	16
57	RNA Misprocessing in C9orf72-Linked Neurodegeneration. <i>Frontiers in Cellular Neuroscience</i> , 2017, 11, .	3.5	32
58	A clinical tool for predicting survival in ALS. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 1361-1367.	2.0	65
59	<scp>ALS</scp>/<scp>FTD</scp>â€associated<scp>FUS</scp> activates<scp>GSK</scp>â€3â€ to disrupt the<scp>VAPB</scp>â€<scp>PTPIP</scp>51 interaction and<scp>ER</scp>â€ mitochondria associations. <i>EMBO Reports</i> , 2016, 17, 1326-1342.	5.3	211
60	Gain of Toxicity from ALS/FTD-Linked Repeat Expansions in C9ORF72 Is Alleviated by Antisense Oligonucleotides Targeting GGGGCC-Containing RNAs. <i>Neuron</i> , 2016, 90, 535-550.	12.8	431
61	Rare genetic variation in UNC13A may modify survival in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2016, 17, 593-599.	2.9	18
62	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	16.3	414
63	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	16.3	199
64	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , 2016, 7, .	14.1	173
65	Reply: The role ofDNAJB2in amyotrophic lateral sclerosis. <i>Brain</i> , 2016, 139, e58-e58.	8.9	0
66	Association of a Locus in the<i>CAMTA1</i>Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2016, 73, 812.	14.3	55
67	The heat shock response plays an important role in TDP-43 clearance: evidence for dysfunction in amyotrophic lateral sclerosis. <i>Brain</i> , 2016, 139, 1417-1432.	8.9	129
68	Maturation and electrophysiological properties of human pluripotent stem cell-derived oligodendrocytes. <i>Stem Cells</i> , 2016, 34, 1040-1053.	3.3	63
69	Lack of association between TDP-43 pathology and tau mis-splicing in Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 37, 45-46.	3.4	8
70	Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. <i>Acta Neuropathologica Communications</i> , 2016, 4, .	5.1	42
71	ALS-FUS pathology revisited: singleton FUS mutations and an unusual case with both a FUS and TARDBP mutation. <i>Acta Neuropathologica Communications</i> , 2015, 3, .	5.1	23
72	U1 snRNP is mislocalized in ALS patient fibroblasts bearing NLS mutations in FUS and is required for motor neuron outgrowth in zebrafish. <i>Nucleic Acids Research</i> , 2015, 43, 3208-3218.	16.2	70

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73	TDP-43 Proteinopathy and ALS: Insights into Disease Mechanisms and Therapeutic Targets. <i>Neurotherapeutics</i> , 2015, 12, 352-363.	6.5	242
74	VCP mutations are not a major cause of familial amyotrophic lateral sclerosis in the UK. <i>Journal of the Neurological Sciences</i> , 2015, 349, 209-213.	1.4	9
75	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. <i>Science</i> , 2015, 347, 1436-1441.	38.2	763
76	Impact of disease, cognitive and behavioural factors on caregiver outcome in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2015, 16, 316-323.	2.9	33
77	Regionality of disease progression predicts prognosis in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2015, 16, 442-447.	2.9	4
78	Genetic analysis of amyotrophic lateral sclerosis in the Slovenian population. <i>Neurobiology of Aging</i> , 2015, 36, 1601.e17-1601.e20.	3.4	10
79	Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. <i>Neurobiology of Aging</i> , 2015, 36, 2006.e1-2006.e9.	3.4	19
80	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. <i>Journal of Neurology</i> , 2015, 262, 1376-1378.	3.4	40
81	The role of <i>TREM2</i> R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. <i>Alzheimer's and Dementia</i> , 2015, 11, 1407-1416.	0.7	153
82	Phosphorylation of C-terminal tyrosine 526 in FUS impairs its nuclear import. <i>Journal of Cell Science</i> , 2015, , .	3.2	24
83	Wild type human TDP-43 potentiates ALS-linked mutant TDP-43 driven progressive motor and cortical neuron degeneration with pathological features of ALS. <i>Acta Neuropathologica Communications</i> , 2015, 3, .	5.1	74
84	Human iPSC-derived motoneurons harbouring TARDBP or C9ORF72 ALS mutations are dysfunctional despite maintaining viability. <i>Nature Communications</i> , 2015, 6, .	14.1	219
85	Executive dysfunction predicts social cognition impairment in amyotrophic lateral sclerosis. <i>Journal of Neurology</i> , 2015, 262, 1681-1690.	3.4	37
86	The CHCHD10 P34S variant is not associated with ALS in a UK cohort of familial and sporadic patients. <i>Neurobiology of Aging</i> , 2015, 36, 2908.e17-2908.e18.	3.4	19
87	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. <i>Acta Neuropathologica Communications</i> , 2015, 3, .	5.1	77
88	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. <i>Neurobiology of Aging</i> , 2015, 36, 1602.e17-1602.e27.	3.4	80
89	An Evaluation of a SVA Retrotransposon in the FUS Promoter as a Transcriptional Regulator and Its Association to ALS. <i>PLoS ONE</i> , 2014, 9, e90833.	2.5	27
90	ERâ€œmitochondria associations are regulated by the VAPBâ€œPTPIP51 interaction and are disrupted by ALS/FTD-associated TDP-43. <i>Nature Communications</i> , 2014, 5, .	14.1	474

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91	A serum microRNA signature for amyotrophic lateral sclerosis reveals convergent RNA processing defects and identifies presymptomatic mutation carriers. <i>Brain</i> , 2014, 137, 2875-2876.	8.9	0
92	Psychological as well as illness factors influence acceptance of non-invasive ventilation (NIV) and gastrostomy in amyotrophic lateral sclerosis (ALS): A prospective population study. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 376-387.	2.9	48
93	Differential roles of the ubiquitin proteasome system (UPS) and autophagy in the clearance of soluble and aggregated TDP-43 species. <i>Journal of Cell Science</i> , 2014, , .	3.2	197
94	Autosomal dominant inheritance of rapidly progressive amyotrophic lateral sclerosis due to a truncation mutation in the fused in sarcoma (FUS) gene. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 557-562.	2.9	11
95	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in <i>C9orf72</i> reveals marked differences in results among 14 laboratories. <i>Journal of Medical Genetics</i> , 2014, 51, 419-424.	3.7	102
96	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2014, 23, 2220-2231.	3.1	110
97	Analysis of amyotrophic lateral sclerosis as a multistep process: a population-based modelling study. <i>Lancet Neurology</i> , The, 2014, 13, 1108-1113.	19.1	312
98	The evaluation of pain in amyotrophic lateral sclerosis: A case controlled observational study. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 520-527.	2.9	57
99	No evidence for shared genetic basis of common variants in multiple sclerosis and amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2014, 23, 1916-1922.	3.1	21
100	Autophagy induction enhances TDP43 turnover and survival in neuronal ALS models. <i>Nature Chemical Biology</i> , 2014, 10, 677-685.	7.3	341
101	<i>C9orf72</i> and <i>UNC13A</i> are shared risk loci for amyotrophic lateral sclerosis and frontotemporal dementia: A genome-wide meta-analysis. <i>Annals of Neurology</i> , 2014, 76, 120-133.	6.6	80
102	Evidence of an environmental effect on survival in ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 528-533.	2.9	15
103	Estimating clinical stage of amyotrophic lateral sclerosis from the ALS Functional Rating Scale. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 279-284.	2.9	112
104	Tar DNA-binding protein-43 (TDP-43) regulates axon growth in vitro and in vivo. <i>Neurobiology of Disease</i> , 2014, 65, 25-34.	5.2	25
105	Allele-Specific Knockdown of ALS-Associated Mutant TDP-43 in Neural Stem Cells Derived from Induced Pluripotent Stem Cells. <i>PLoS ONE</i> , 2014, 9, e91269.	2.5	38
106	Residual association at <i>C9orf72</i> suggests an alternative amyotrophic lateral sclerosis-causing hexanucleotide repeat. <i>Neurobiology of Aging</i> , 2013, 34, 2234.e1-2234.e7.	3.4	21
107	Neuromuscular disease: new insights and avenues for therapy. <i>Lancet Neurology</i> , The, 2013, 12, 13-15.	19.1	2
108	Association studies indicate that protein disulfide isomerase is a risk factor in amyotrophic lateral sclerosis. <i>Free Radical Biology and Medicine</i> , 2013, 58, 81-86.	3.0	43

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109	H63D polymorphism in HFE is not associated with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2013, 34, 1517.e5-1517.e7.	3.4	18
110	pNfH is a promising biomarker for ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 146-149.	2.9	64
111	ALS-linked TDP-43 mutations produce aberrant RNA splicing and adult-onset motor neuron disease without aggregation or loss of nuclear TDP-43. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, .	7.7	358
112	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. <i>Nature</i> , 2013, 495, 467-473.	40.1	1,150
113	Comment on "Drug Screening for ALS Using Patient-Specific Induced Pluripotent Stem Cells". <i>Science Translational Medicine</i> , 2013, 5, .	13.1	7
114	Loss and gain of Drosophila TDP-43 impair synaptic efficacy and motor control leading to age-related neurodegeneration by loss-of-function phenotypes. <i>Human Molecular Genetics</i> , 2013, 22, 1539-1557.	3.1	108
115	Is language impairment more common than executive dysfunction in amyotrophic lateral sclerosis?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 494-498.	2.0	126
116	ALS mutant FUS disrupts nuclear localization and sequesters wild-type FUS within cytoplasmic stress granules. <i>Human Molecular Genetics</i> , 2013, 22, 2676-2688.	3.1	189
117	Drosophila TDP-43 dysfunction in glia and muscle cells cause cytological and behavioural phenotypes that characterize ALS and FTL. <i>Human Molecular Genetics</i> , 2013, 22, 3883-3893.	3.1	65
118	Astrocyte pathology and the absence of non-cell autonomy in an induced pluripotent stem cell model of TDP-43 proteinopathy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 4697-4702.	7.7	279
119	Unpicking neurodegeneration in a dish with human pluripotent stem cells. <i>Cell Cycle</i> , 2013, 12, 2339-2340.	3.2	2
120	Modelling C9ORF72 hexanucleotide repeat expansion in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Acta Neuropathologica</i> , 2013, 127, 377-389.	7.9	41
121	Downregulation of MicroRNA-9 in iPSC-Derived Neurons of FTD/ALS Patients with TDP-43 Mutations. <i>PLoS ONE</i> , 2013, 8, e76055.	2.5	105
122	Amyotrophic lateral sclerosis-associated mutant VAPBP56S perturbs calcium homeostasis to disrupt axonal transport of mitochondria. <i>Human Molecular Genetics</i> , 2012, 21, 1979-1988.	3.1	114
123	VAPB interacts with the mitochondrial protein PTPIP51 to regulate calcium homeostasis. <i>Human Molecular Genetics</i> , 2012, 21, 1299-1311.	3.1	432
124	No evidence for a large difference in ALS frequency in populations of African and European origin: A population based study in inner city London. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 66-68.	2.3	12
125	The Role of Variation at AÎ²PP, PSEN1, PSEN2, and MAPT in Late Onset Alzheimer's Disease. <i>Journal of Alzheimer's Disease</i> , 2012, 28, 377-387.	2.7	50
126	Polymorphisms in the GluR2 gene are not associated with amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2012, 33, 418-420.	3.4	9

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127	Mutation analysis of VCP in British familial and sporadic amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , 2012, 33, 2721.e1-2721.e2.	3.4	16
128	Screening for OPTN mutations in a cohort of British amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , 2012, 33, 2948.e15-2948.e17.	3.4	17
129	Mutant induced pluripotent stem cell lines recapitulate aspects of TDP-43 proteinopathies and reveal cell-specific vulnerability. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 5803-5808.	7.7	276
130	A proposed staging system for amyotrophic lateral sclerosis. <i>Brain</i> , 2012, 135, 847-852.	8.9	334
131	Widespread binding of FUS along nascent RNA regulates alternative splicing in the brain. <i>Scientific Reports</i> , 2012, 2, .	3.7	213
132	Eating-derived pleasure in amyotrophic lateral sclerosis as a predictor of non-oral feeding. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 555-559.	2.3	18
133	Mapping of Gene Expression Reveals CYP27A1 as a Susceptibility Gene for Sporadic ALS. <i>PLoS ONE</i> , 2012, 7, e35333.	2.5	46
134	Amyotrophic lateral sclerosis and other disorders of the lower motor neuron. , 2012, , 136-147.		0
135	An MND/ALS phenotype associated with <i>C9orf72</i> repeat expansion: Abundant p62 ⁺ positive, TDP ⁺ negative inclusions in cerebral cortex, hippocampus and cerebellum but without associated cognitive decline. <i>Neuropathology</i> , 2012, 32, 505-514.	1.3	110
136	Mixed tau, TDP-43 and p62 pathology in FTL associated with a C9ORF72 repeat expansion and p.Ala239Thr MAPT (tau) variant. <i>Acta Neuropathologica</i> , 2012, 125, 303-310.	7.9	69
137	Overexpression of human wild-type FUS causes progressive motor neuron degeneration in an age- and dose-dependent fashion. <i>Acta Neuropathologica</i> , 2012, 125, 273-288.	7.9	216
138	The C9ORF72 expansion mutation is a common cause of ALS+/FTD in Europe and has a single founder. <i>European Journal of Human Genetics</i> , 2012, 21, 102-108.	3.1	185
139	Combination of neurofilament heavy chain and complement C3 as CSF biomarkers for ALS. <i>Journal of Neurochemistry</i> , 2011, 117, 528-537.	4.0	123
140	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. <i>Nature Genetics</i> , 2011, 43, 429-435.	16.3	1,637
141	Characterizing the RNA targets and position-dependent splicing regulation by TDP-43. <i>Nature Neuroscience</i> , 2011, 14, 452-458.	12.4	874
142	Endosomal accumulation of APP in wobbler motor neurons reflects impaired vesicle trafficking: Implications for human motor neuron disease. <i>BMC Neuroscience</i> , 2011, 12, .	2.2	35
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