Kevin Talbot

List of Publications by Year
in descending order

250 papers	19,259 citations	8181 76 h-index	14759 127 g-index
260 all docs	260 docs citations	260 times ranked	19717 citing authors

#	Article	IF	Citations
1	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	10.2	1,039
2	Mutant small heat-shock protein 27 causes axonal Charcot-Marie-Tooth disease and distal hereditary motor neuropathy. Nature Genetics, 2004, 36, 602-606.	21.4	541
3	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
4	Controversies and priorities in amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 310-322.	10.2	454
5	Transgenics, toxicity and therapeutics in rodent models of mutant SOD1-mediated familial ALS. Progress in Neurobiology, 2008, 85, 94-134.	5.7	435
6	Neurofilament light chain. Neurology, 2015, 84, 2247-2257.	1.1	412
7	Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. Nature Genetics, 2004, 36, 597-601.	21.4	395
8	Biomarkers in amyotrophic lateral sclerosis. Lancet Neurology, The, 2009, 8, 94-109.	10.2	391
9	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
10	Selective vulnerability of motor neurons and dissociation of pre- and post-synaptic pathology at the neuromuscular junction in mouse models of spinal muscular atrophy. Human Molecular Genetics, 2008, 17, 949-962.	2.9	333
11	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. American Journal of Human Genetics, 2016, 98, 500-513.	6.2	333
12	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
13	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
14	Extracellular vesicles in neurodegenerative disease â€" pathogenesis to biomarkers. Nature Reviews Neurology, 2016, 12, 346-357.	10.1	299
15	ER Stress and Autophagic Perturbations Lead to Elevated Extracellular Î±-Synuclein in GBA-N370S Parkinson's iPSC-Derived Dopamine Neurons. Stem Cell Reports, 2016, 6, 342-356.	4.8	279
16	A proposal for new diagnostic criteria for ALS. Clinical Neurophysiology, 2020, 131, 1975-1978.	1.5	268
17	Corpus callosum involvement is a consistent feature of amyotrophic lateral sclerosis. Neurology, 2010, 75, 1645-1652.	1.1	257
18	Deletions in the survival motor neuron gene on 5q13 in autosomal recessive spinal muscular atrophy. Human Molecular Genetics, 1995, 4, 631-634.	2.9	233

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19	Integration of structural and functional magnetic resonance imaging in amyotrophic lateral sclerosis. Brain, 2011, 134, 3470-3479.	7.6	229
20	The influence of age and gender on motor and non-motor features of early Parkinson's disease: Initial findings from the Oxford Parkinson Disease Center (OPDC) discovery cohort. Parkinsonism and Related Disorders, 2014, 20, 99-105.	2.2	223
21	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
22	Alternative Splicing Events Are a Late Feature of Pathology in a Mouse Model of Spinal Muscular Atrophy. PLoS Genetics, 2009, 5, e1000773.	3.5	210
23	C9orf72 Hexanucleotide Expansions Are Associated with Altered Endoplasmic Reticulum Calcium Homeostasis and Stress Granule Formation in Induced Pluripotent Stem Cell-Derived Neurons from Patients with Amyotrophic Lateral Sclerosis and Frontotemporal Dementia. Stem Cells, 2016, 34, 2063-2078.	3.2	195
24	Juvenile ALS with basophilic inclusions is a FUS proteinopathy with FUS mutations. Neurology, 2010, 75, 611-618.	1.1	178
25	Advances in therapy for spinal muscular atrophy: promises and challenges. Nature Reviews Neurology, 2018, 14, 214-224.	10.1	174
26	Compound Heterozygosity for Loss-of-Function Lysyl-tRNA Synthetase Mutations in a Patient with Peripheral Neuropathy. American Journal of Human Genetics, 2010, 87, 560-566.	6.2	169
27	Systemic peptide-mediated oligonucleotide therapy improves long-term survival in spinal muscular atrophy. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 10962-10967.	7.1	159
28	Autoimmune disease preceding amyotrophic lateral sclerosis. Neurology, 2013, 81, 1222-1225.	1.1	156
29	Improving clinical trial outcomes in amyotrophic lateral sclerosis. Nature Reviews Neurology, 2021, 17, 104-118.	10.1	152
30	Widespread grey matter pathology dominates the longitudinal cerebral MRI and clinical landscape of amyotrophic lateral sclerosis. Brain, 2014, 137, 2546-2555.	7.6	151
31	REM sleep behaviour disorder is associated with worse quality of life and other non-motor features in early Parkinson's disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 560-566.	1.9	150
32	Next generation sequencing for molecular diagnosis of neurological disorders using ataxias as a model. Brain, 2013, 136, 3106-3118.	7.6	146
33	Missense mutation clustering in the survival motor neuron gene: a role for a conserved tyrosine and glycine rich region of the protein in RNA metabolism?. Human Molecular Genetics, 1997, 6, 497-500.	2.9	145
34	Genetics of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2007, 16, R233-R242.	2.9	145
35	A mutation in the small heat-shock protein HSPB1 leading to distal hereditary motor neuronopathy disrupts neurofilament assembly and the axonal transport of specific cellular cargoes. Human Molecular Genetics, 2006, 15, 347-354.	2.9	138
36	The clinical landscape for SMA in a new therapeutic era. Gene Therapy, 2017, 24, 529-533.	4.5	133

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37	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
38	C9orf72 and RAB7L1 regulate vesicle trafficking in amyotrophic lateral sclerosis and frontotemporal dementia. Brain, 2017, 140, 887-897.	7.6	126
39	Predictors of cognitive impairment in an early stage Parkinson's disease cohort. Movement Disorders, 2014, 29, 351-359.	3.9	124
40	Investigation of white matter pathology in ALS and PLS using tractâ€based spatial statistics. Human Brain Mapping, 2009, 30, 615-624.	3.6	123
41	Review: Neuromuscular synaptic vulnerability in motor neurone disease: amyotrophic lateral sclerosis and spinal muscular atrophy. Neuropathology and Applied Neurobiology, 2010, 36, 133-156.	3.2	123
42	Systematic review of methodology used in ultrasound studies aimed at creating charts of fetal size. BJOG: an International Journal of Obstetrics and Gynaecology, 2012, 119, 1425-1439.	2.3	119
43	Lithium in patients with amyotrophic lateral sclerosis (LiCALS): a phase 3 multicentre, randomised, double-blind, placebo-controlled trial. Lancet Neurology, The, 2013, 12, 339-345.	10.2	118
44	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
45	<scp>CSF</scp> neurofilament light chain reflects corticospinal tract degeneration in <scp>ALS</scp>. Annals of Clinical and Translational Neurology, 2015, 2, 748-755.	3.7	118
46	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	8.2	117
47	Motor neuron disease: THE BARE ESSENTIALS. Practical Neurology, 2009, 9, 303-309.	1.1	115
48	Defective cholesterol metabolism in amyotrophic lateral sclerosis. Journal of Lipid Research, 2017, 58, 267-278.	4.2	115
49	Concordance between site of onset and limb dominance in amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 853-854.	1.9	114
50	The contribution of mouse models to understanding the pathogenesis of spinal muscular atrophy. DMM Disease Models and Mechanisms, 2011, 4, 457-467.	2.4	113
51	Oculomotor Dysfunction in Amyotrophic Lateral Sclerosis. Archives of Neurology, 2011, 68, 857.	4.5	112
52	Identification of distinct circulating exosomes in Parkinson's disease. Annals of Clinical and Translational Neurology, 2015, 2, 353-361.	3.7	111
53	Heterotopic pregnancy. Journal of Obstetrics and Gynaecology, 2011, 31, 7-12.	0.9	109
54	Charcot-Marie-Toothâ€“Linked Mutant GARS Is Toxic to Peripheral Neurons Independent of Wild-Type GARS Levels. PLoS Genetics, 2011, 7, e1002399.	3.5	109

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55	Cerebrospinal fluid macrophage biomarkers in amyotrophic lateral sclerosis. Annals of Neurology, 2018, 83, 258-268.	5.3	107
56	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106
57	SMN deficiency disrupts brain development in a mouse model of severe spinal muscular atrophy. Human Molecular Genetics, 2010, 19, 4216-4228.	2.9	105
58	Genetic screening in sporadic ALS and FTD. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 1042-1044.	1.9	105
59	A Loss-of-Function Variant in the Human Histidyl-tRNA Synthetase (HARS) Gene is Neurotoxic In Vivo. Human Mutation, 2013, 34, 191-199.	2.5	104
60	Towards a TDP-43-Based Biomarker for ALS and FTLD. Molecular Neurobiology, 2018, 55, 7789-7801.	4.0	100
61	The two-year progression of structural and functional cerebral MRI in amyotrophic lateral sclerosis. NeuroImage: Clinical, 2018, 17, 953-961.	2.7	100
62	Pre-symptomatic development of lower motor neuron connectivity in a mouse model of severe spinal muscular atrophy. Human Molecular Genetics, 2010, 19, 420-433.	2.9	98
63	Pathogenesis of FUS-associated ALS and FTD: insights from rodent models. Acta Neuropathologica Communications, 2016, 4, 99.	5.2	97
64	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
65	Advances in motor neurone disease. Journal of the Royal Society of Medicine, 2014, 107, 14-21.	2.0	93
66	HDAC6 is a therapeutic target in mutant GARS-induced Charcot-Marie-Tooth disease. Brain, 2018, 141, 673-687.	7.6	93
67	Motor neurone disease. Postgraduate Medical Journal, 2002, 78, 513-519.	1.8	92
68	An ENU-induced mutation in mouse glycyl-tRNA synthetase (GARS) causes peripheral sensory and motor phenotypes creating a model of Charcot-Marie-Tooth type 2D peripheral neuropathy. DMM Disease Models and Mechanisms, 2009, 2, 359-373.	2.4	91
69	A Recurrent loss-of-function alanyl-tRNA synthetase (AARSâ€‰) mutation in patients with charcot-marie-tooth disease type 2N (CMT2N). Human Mutation, 2012, 33, 244-253.	2.5	90
70	Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. Human Molecular Genetics, 2011, 20, 4334-4344.	2.9	89
71	The diagnostic pathway and prognosis in bulbar-onset amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2010, 294, 81-85.	0.6	87
72	Therapeutic strategies for spinal muscular atrophy: SMN and beyond. DMM Disease Models and Mechanisms, 2017, 10, 943-954.	2.4	87

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73	Impairment of Mitochondrial Calcium Buffering Links Mutations in C9ORF72 and TARDBP in iPS-Derived Motor Neurons from Patients with ALS/FTD. Stem Cell Reports, 2020, 14, 892-908.	4.8	86
74	Safety and efficacy of diaphragm pacing in patients with respiratory insufficiency due to amyotrophic lateral sclerosis (DiPALS): a multicentre, open-label, randomised controlled trial. Lancet Neurology, The, 2015, 14, 883-892.	10.2	85
75	Mimics and chameleons in motor neurone disease. Practical Neurology, 2013, 13, 153-164.	1.1	84
76	Comprehensive morphometry of subcortical grey matter structures in earlyâ€stage Parkinson's disease. Human Brain Mapping, 2014, 35, 1681-1690.	3.6	84
77	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. Brain, 2019, 142, 586-605.	7.6	84
78	Reduction in excess daytime sleepiness by modafinil in patients with myotonic dystrophy. Neuromuscular Disorders, 2003, 13, 357-364.	0.6	83
79	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. Acta Neuropathologica Communications, 2017, 5, 13.	5.2	83
80	Increased functional connectivity common to symptomatic amyotrophic lateral sclerosis and those at genetic risk. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 580-588.	1.9	82
81	Recent advances in the genetics of amyotrophic lateral sclerosis and frontotemporal dementia: common pathways in neurodegenerative disease. Human Molecular Genetics, 2006, 15, R182-R187.	2.9	81
82	Vascular <scp>D</scp>efects and <scp>S</scp>pinal <scp>C</scp>ord <scp>H</scp>ypoxia in <scp>S</scp>pinal <scp>M</scp>uscular <scp>A</scp>trophy. Annals of Neurology, 2016, 79, 217-230.	5.3	79
83	Non-neuronal cells in amyotrophic lateral sclerosis â€" from pathogenesis to biomarkers. Nature Reviews Neurology, 2021, 17, 333-348.	10.1	78
84	Gene deletions in spinal muscular atrophy.. Journal of Medical Genetics, 1996, 33, 93-96.	3.2	77
85	Neuromuscular junction maturation defects precede impaired lower motor neuron connectivity in Charcot-Marie-Tooth type 2D mice. Human Molecular Genetics, 2014, 23, 2639-2650.	2.9	75
86	Severe childhood SMA and axonal CMT due to anticodon binding domain mutations in the GARS gene. Neurology, 2006, 67, 1710-1712.	1.1	72
87	GARS axonopathy: not every neuron's cup of tRNA. Trends in Neurosciences, 2010, 33, 59-66.	8.6	70
88	Spinal Muscular Atrophy. Seminars in Neurology, 2001, 21, 189-198.	1.4	68
89	Vitamin E deficiency induced neurological disease in common variable immunodeficiency: two cases and a review of the literature of vitamin E deficiency. Clinical Immunology, 2004, 112, 24-29.	3.2	67
90	Axonal TDP-43 condensates drive neuromuscular junction disruption through inhibition of local synthesis of nuclear encoded mitochondrial proteins. Nature Communications, 2021, 12, 6914.	12.8	67

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91	CSF chitinase proteins in amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1215-1220.	1.9	66
92	The ALSFRS as an outcome measure in therapeutic trials and its relationship to symptom onset. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 414-425.	1.7	65
93	Eye-tracking in amyotrophic lateral sclerosis: A longitudinal study of saccadic and cognitive tasks. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 101-111.	1.7	65
94	Characterization of a gene encoding survival motor neuron (SMN)-related protein, a constituent of the spliceosome complex. Human Molecular Genetics, 1998, 7, 2149-2156.	2.9	64
95	Survival motor neuron deficiency enhances progression in an amyotrophic lateral sclerosis mouse model. Neurobiology of Disease, 2009, 34, 511-517.	4.4	62
96	Cardiovascular fitness as a risk factor for amyotrophic lateral sclerosis: indirect evidence from record linkage study: Table 1. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 395-398.	1.9	62
97	Chondrolectin affects cell survival and neuronal outgrowth in in vitro and in vivo models of spinal muscular atrophy. Human Molecular Genetics, 2014, 23, 855-869.	2.9	62
98	Single-copy expression of an amyotrophic lateral sclerosis-linked TDP-43 mutation (M337V) in BAC transgenic mice leads to altered stress granule dynamics and progressive motor dysfunction. Neurobiology of Disease, 2019, 121, 148-162.	4.4	62
99	Trk receptor signaling and sensory neuron fate are perturbed in human neuropathy caused by Gars mutations. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E3324-E3333.	7.1	61
100	Spinal muscular atrophy: antisense oligonucleotide therapy opens the door to an integrated therapeutic landscape. Human Molecular Genetics, 2017, 26, R151-R159.	2.9	61
101	Fractional Anisotropy in the Posterior Limb of the Internal Capsule and Prognosis in Amyotrophic Lateral Sclerosis. Archives of Neurology, 2012, 69, 1493.	4.5	60
102	Treatment of vasculitic peripheral neuropathy: a retrospective analysis of outcome. QJM - Monthly Journal of the Association of Physicians, 2006, 100, 41-51.	0.5	59
103	Myelin imaging in amyotrophic and primary lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 562-573.	1.7	59
104	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
105	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
106	Altered cortical betaâ€band oscillations reflect motor system degeneration in amyotrophic lateral sclerosis. Human Brain Mapping, 2017, 38, 237-254.	3.6	58
107	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
108	TDP-43 expression in mouse models of amyotrophic lateral sclerosis and spinal muscular atrophy. BMC Neuroscience, 2008, 9, 104.	1.9	55

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109	Dismutase-competent SOD1 mutant accumulation in myelinating Schwann cells is not detrimental to normal or transgenic ALS model mice. Human Molecular Genetics, 2010, 19, 815-824.	2.9	52
110	Mice Carrying ALS Mutant TDP-43, but Not Mutant FUS, Display InÂVivo Defects in Axonal Transport of Signaling Endosomes. Cell Reports, 2020, 30, 3655-3662.e2.	6.4	51
111	Whole-brain magnetic resonance spectroscopic imaging measures are related to disability in ALS. Neurology, 2013, 80, 610-615.	1.1	50
112	Dominant, toxic gain-of-function mutations ingarslead to non-cell autonomous neuropathology. Human Molecular Genetics, 2015, 24, 4397-4406.	2.9	50
113	HspB8 mutation causing hereditary distal motor neuropathy impairs lysosomal delivery of autophagosomes. Journal of Neurochemistry, 2011, 119, 1155-1161.	3.9	49
114	The longitudinal cerebrospinal fluid metabolomic profile of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 456-463.	1.7	49
115	Psychiatric disorders prior to amyotrophic lateral sclerosis. Annals of Neurology, 2016, 80, 935-938.	5.3	49
116	Head and other physical trauma requiring hospitalisation is not a significant risk factor in the development of ALS. Journal of the Neurological Sciences, 2010, 288, 45-48.	0.6	48
117	TARDBP pathogenic mutations increase cytoplasmic translocation of TDP-43 and cause reduction of endoplasmic reticulum Ca2+ signaling in motor neurons. Neurobiology of Disease, 2015, 75, 64-77.	4.4	45
118	Genetic compendium of 1511 human brains available through the UK Medical Research Council Brain Banks Network Resource. Genome Research, 2017, 27, 165-173.	5.5	44
119	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 666-673.	1.9	43
120	The Role of Mitochondrial Dysfunction and ER Stress in TDP-43 and C9ORF72 ALS. Frontiers in Cellular Neuroscience, 2021, 15, 653688.	3.7	43
121	The molecular genetics of non-ALS motor neuron diseases. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 986-1000.	3.8	41
122	Asymmetrical late onset motor neuropathy associated with a novel mutation in the small heat shock protein HSPB1 (HSP27). Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 461-463.	1.9	41
123	July 2017 ENCALS statement on edaravone. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 471-474.	1.7	41
124	Overexpression of survival motor neuron improves neuromuscular function and motor neuron survival in mutant SOD1 mice. Neurobiology of Aging, 2014, 35, 906-915.	3.1	39
125	Regional thalamic MRI as a marker of widespread cortical pathology and progressive frontotemporal involvement in amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1250-1258.	1.9	39
126	Correction of amyotrophic lateral sclerosis related phenotypes in induced pluripotent stem cell-derived motor neurons carrying a hexanucleotide expansion mutation in C9orf72 by CRISPR/Cas9 genome editing using homology-directed repair. Human Molecular Genetics, 2020, 29, 2200-2217.	2.9	39

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127	Human stem cell models of neurodegeneration: From basic science of amyotrophic lateral sclerosis to clinical translation. Cell Stem Cell, 2022, 29, 11-35.	11.1	39
128	Neurodegeneration in SCA14 is associated with increased PKCÎ³ kinase activity, mislocalization and aggregation. Acta Neuropathologica Communications, 2018, 6, 99.	5.2	37
129	Impaired corticomuscular and interhemispheric cortical beta oscillation coupling in amyotrophic lateral sclerosis. Clinical Neurophysiology, 2018, 129, 1479-1489.	1.5	36
130	UFLCâ€Derived CSF Extracellular Vesicle Origin and Proteome. Proteomics, 2018, 18, e1800257.	2.2	36
131	Amyotrophic lateral sclerosis: the complex path to precision medicine. Journal of Neurology, 2018, 265, 2454-2462.	3.6	36
132	Deep phenotyping of peripheral tissue facilitates mechanistic disease stratification in sporadic Parkinsonâ€™s disease. Progress in Neurobiology, 2020, 187, 101772.	5.7	35
133	Identification of a potential non-coding RNA biomarker signature for amyotrophic lateral sclerosis. Brain Communications, 2020, 2, fcaa053.	3.3	34
134	Spinal muscular atrophy. Journal of Inherited Metabolic Disease, 1999, 22, 545-554.	3.6	33
135	Functional vitamin B12 deficiency. Practical Neurology, 2009, 9, 37-45.	1.1	33
136	T₂-Weighted MRI Detects Presymptomatic Pathology in the SOD1 Mouse Model of ALS. Journal of Cerebral Blood Flow and Metabolism, 2014, 34, 785-793.	4.3	32
137	Amyotrophic lateral sclerosis: cell vulnerability or system vulnerability?. Journal of Anatomy, 2014, 224, 45-51.	1.5	32
138	Objectively Monitoring Amyotrophic Lateral Sclerosis Patient Symptoms During Clinical Trials With Sensors: Observational Study. JMIR MHealth and UHealth, 2019, 7, e13433.	3.7	32
139	Development of a patient reported outcome measure for fatigue in motor neurone disease: the Neurological Fatigue Index (NFI-MND). Health and Quality of Life Outcomes, 2011, 9, 101.	2.4	31
140	The impact of fatigue and psychosocial variables on quality of life for patients with motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 537-545.	1.7	31
141	Cerebrovascular injury as a risk factor for amyotrophic lateral sclerosis: TableÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 244-246.	1.9	31
142	ADCY5-related dyskinesia presenting as familial myoclonus-dystonia. Neurogenetics, 2017, 18, 111-117.	1.4	31
143	Quantitative FLAIR MRI in Amyotrophic Lateral Sclerosis. Academic Radiology, 2017, 24, 1187-1194.	2.5	31
144	Vasoactive intestinal peptide, but not pituitary adenylate cyclase-activating peptide, modulates the responsiveness of the gonadotroph to LHRH in man. Journal of Endocrinology, 1993, 137, 529-532.	2.6	30

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145	An ALS-linked mutation in TDP-43 disrupts normal protein interactions in the motor neuron response to oxidative stress. Neurobiology of Disease, 2020, 144, 105050.	4.4	30
146	Characterisation of novel point mutations in the survival motor neuron gene SMN , in three patients with SMA. Human Genetics, 2001, 108, 356-357.	3.8	29
147	The role of RNA processing in the pathogenesis of motor neuron degeneration. Expert Reviews in Molecular Medicine, 2010, 12, e21.	3.9	29
148	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029.	3.3	29
149	Gene conversion at the SMN locus in autosomal recessive spinal muscular atrophy does not predict a mild phenotype. Neuromuscular Disorders, 1997, 7, 198-201.	0.6	28
150	Crystal structure of human wildtype and S581L-mutant glycyl-tRNA synthetase, an enzyme underlying distal spinal muscular atrophy. FEBS Letters, 2007, 581, 2959-2964.	2.8	28
151	TARDBP in amyotrophic lateral sclerosis: identification of a novel variant but absence of copy number variation. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 1283-1285.	1.9	28
152	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
153	Normal d-dimer levels do not exclude the diagnosis of cerebral venous sinus thrombosis. Journal of Neurology, 2002, 249, 1603-1604.	3.6	27
154	Whole-exome sequencing of 228 patients with sporadic Parkinsonâ€™s disease. Scientific Reports, 2017, 7, 41188.	3.3	27
155	The relationships between symptoms, disability, perceived health and quality of life in amyotrophic lateral sclerosis/motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 317-327.	1.7	27
156	Quantitative patterns of motor cortex proteinopathy across ALS genotypes. Acta Neuropathologica Communications, 2020, 8, 98.	5.2	27
157	CSF extracellular vesicle proteomics demonstrates altered protein homeostasis in amyotrophic lateral sclerosis. Clinical Proteomics, 2020, 17, 31.	2.1	27
158	A case of celiac disease mimicking amyotrophic lateral sclerosis. Nature Clinical Practice Neurology, 2007, 3, 581-584.	2.5	26
159	Novel SPG11 mutations in Asian kindreds and disruption of spatacsin function in the zebrafish. Neurogenetics, 2010, 11, 379-389.	1.4	26
160	Increased cerebral functional connectivity in ALS. Neurology, 2018, 90, e1418-e1424.	1.1	26
161	Cerebellar tract alterations in PLS and ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 281-284.	1.7	26
162	CSF chitinases before and after symptom onset in amyotrophic lateral sclerosis. Annals of Clinical and Translational Neurology, 2020, 7, 1296-1306.	3.7	26

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163	Detection and quantification of novel Câ€terminal TDPâ€43 fragments in ALSâ€TDP. Brain Pathology, 2021, 31, e12923.	4.1	26
164	Higher blood high density lipoprotein and apolipoprotein A1 levels are associated with reduced risk of developing amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 75-81.	1.9	24
165	FTLD-ALS of TDP-43 type and SCA2 in a family with a full ataxin-2 polyglutamine expansion. Acta Neuropathologica, 2014, 128, 597-604.	7.7	23
166	Familial versus sporadic amyotrophic lateral sclerosis--a false dichotomy?. Brain, 2011, 134, 3429-3434.	7.6	22
167	An Eye-Tracking Version of the Trail-Making Test. PLoS ONE, 2013, 8, e84061.	2.5	21
168	A multicentre evaluation of oropharyngeal secretion management practices in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 1-9.	1.7	20
169	Frequency and signature of somatic variants in 1461 human brain exomes. Genetics in Medicine, 2019, 21, 904-912.	2.4	20
170	Murray Valley encephalitis in an adult traveller complicated by long-term flaccid paralysis: case report and review of the literature. Transactions of the Royal Society of Tropical Medicine and Hygiene, 2007, 101, 284-288.	1.8	19
171	Neuronal over-expression of Oxr1 is protective against ALS-associated mutant TDP-43 mislocalisation in motor neurons and neuromuscular defects in vivo. Human Molecular Genetics, 2019, 28, 3584-3599.	2.9	19
172	Primary lateral sclerosis: diagnosis and management. Practical Neurology, 2020, 20, 262-269.	1.1	19
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