

Kevin Talbot

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/234328/publications.pdf>

Version: 2024-02-01

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	Higher blood high density lipoprotein and apolipoprotein A1 levels are associated with reduced risk of developing amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 75-81.	1.9	24
2	Human stem cell models of neurodegeneration: From basic science of amyotrophic lateral sclerosis to clinical translation. <i>Cell Stem Cell</i> , 2022, 29, 11-35.	11.1	39
3	Genetic testing in motor neurone disease. <i>Practical Neurology</i> , 2022, 22, 107-116.	1.1	17
4	Modeling seeding and neuroanatomic spread of pathology in amyotrophic lateral sclerosis. <i>NeuroImage</i> , 2022, 251, 118968.	4.2	5
5	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. <i>Brain Communications</i> , 2022, 4, fcac029.	3.3	29
6	A case of SOD1 deficiency: implications for clinical trials. <i>Brain</i> , 2022, 145, 805-806.	7.6	3
7	Hyperexcitability in young iPSC-derived C9ORF72 mutant motor neurons is associated with increased intracellular calcium release. <i>Scientific Reports</i> , 2022, 12, 7378.	3.3	6
8	Creatine kinase and prognosis in amyotrophic lateral sclerosis: a literature review and multi-centre cohort analysis. <i>Journal of Neurology</i> , 2022, 269, 5395-5404.	3.6	6
9	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , 2021, 109, 448-460.e4.	8.1	56
10	Detection and quantification of novel C-terminal TDP ⁴³ fragments in ALS ^{TDP} . <i>Brain Pathology</i> , 2021, 31, e12923.	4.1	26
11	Improving clinical trial outcomes in amyotrophic lateral sclerosis. <i>Nature Reviews Neurology</i> , 2021, 17, 104-118.	10.1	152
12	Motor Neuron Disease Register for England, Wales and Northern Ireland TM an analysis of incidence in England. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021, 22, 86-93.	1.7	10
13	A fine balance between Prpf19 and Exoc7 in achieving degradation of aggregated protein and suppression of cell death in spinocerebellar ataxia type 3. <i>Cell Death and Disease</i> , 2021, 12, 136.	6.3	8
14	Targeting the 5' untranslated region of SMN2 as a therapeutic strategy for spinal muscular atrophy. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 23, 731-742.	5.1	3
15	Network Analysis of the CSF Proteome Characterizes Convergent Pathways of Cellular Dysfunction in ALS. <i>Frontiers in Neuroscience</i> , 2021, 15, 642324.	2.8	6
16	Isolated homozygous R217X OPTN mutation causes knock-out of functional C-terminal optineurin domains and associated oligodendroglialopathy-dominant ALS ^{TDP} . <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 1022-1024.	1.9	2
17	Non-neuronal cells in amyotrophic lateral sclerosis TM from pathogenesis to biomarkers. <i>Nature Reviews Neurology</i> , 2021, 17, 333-348.	10.1	78
18	The Role of Mitochondrial Dysfunction and ER Stress in TDP-43 and C9ORF72 ALS. <i>Frontiers in Cellular Neuroscience</i> , 2021, 15, 653688.	3.7	43

#	ARTICLE	IF	CITATIONS
19	Axonal TDP-43 condensates drive neuromuscular junction disruption through inhibition of local synthesis of nuclear encoded mitochondrial proteins. <i>Nature Communications</i> , 2021, 12, 6914.	12.8	67
20	An ALS-linked mutation in TDP-43 disrupts normal protein interactions in the motor neuron response to oxidative stress. <i>Neurobiology of Disease</i> , 2020, 144, 105050.	4.4	30
21	Amyotrophic Lateral Sclerosis: network vulnerability and monosynaptic connections. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 906-906.	1.9	0
22	Quantitative patterns of motor cortex proteinopathy across ALS genotypes. <i>Acta Neuropathologica Communications</i> , 2020, 8, 98.	5.2	27
23	CSF chitinases before and after symptom onset in amyotrophic lateral sclerosis. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1296-1306.	3.7	26
24	Amyotrophic lateral sclerosis with a heterozygous D91A SOD1 variant and classical ALS-TDP neuropathology. <i>Neurology</i> , 2020, 95, 595-596.	1.1	9
25	CSF extracellular vesicle proteomics demonstrates altered protein homeostasis in amyotrophic lateral sclerosis. <i>Clinical Proteomics</i> , 2020, 17, 31.	2.1	27
26	Neurotrophic Properties of C-Terminal Domain of the Heavy Chain of Tetanus Toxin on Motor Neuron Disease. <i>Toxins</i> , 2020, 12, 666.	3.4	2
27	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.	1.7	12
28	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. <i>Human Molecular Genetics</i> , 2020, 29, 2200-2217.	2.9	39
29	Mice Carrying ALS Mutant TDP-43, but Not Mutant FUS, Display In Vivo Defects in Axonal Transport of Signaling Endosomes. <i>Cell Reports</i> , 2020, 30, 3655-3662.e2.	6.4	51
30	Primary lateral sclerosis: diagnosis and management. <i>Practical Neurology</i> , 2020, 20, 262-269.	1.1	19
31	Measuring quality of life in ALS/MND: validation of the WHOQOL-BREF. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 364-372.	1.7	5
32	Identification of a potential non-coding RNA biomarker signature for amyotrophic lateral sclerosis. <i>Brain Communications</i> , 2020, 2, fcaa053.	3.3	34
33	Regional callosal integrity and bilaterality of limb weakness in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 396-402.	1.7	13
34	Deep phenotyping of peripheral tissue facilitates mechanistic disease stratification in sporadic Parkinson's disease. <i>Progress in Neurobiology</i> , 2020, 187, 101772.	5.7	35
35	Impairment of Mitochondrial Calcium Buffering Links Mutations in C9ORF72 and TARDBP in iPS-Derived Motor Neurons from Patients with ALS/FTD. <i>Stem Cell Reports</i> , 2020, 14, 892-908.	4.8	86
36	A proposal for new diagnostic criteria for ALS. <i>Clinical Neurophysiology</i> , 2020, 131, 1975-1978.	1.5	268

#	ARTICLE	IF	CITATIONS
37	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. <i>Journal of Clinical Investigation</i> , 2020, 130, 6080-6092.	8.2	117
38	Development of LNA Gapmer Oligonucleotide-Based Therapy for ALS/FTD Caused by the C9orf72 Repeat Expansion. <i>Methods in Molecular Biology</i> , 2020, 2176, 185-208.	0.9	1
39	Tracheostomy in motor neurone disease. <i>Practical Neurology</i> , 2019, 19, 467-475.	1.1	15
40	Wrangling RNA: Antisense oligonucleotides for neurological disorders. <i>Science Translational Medicine</i> , 2019, 11, .	12.4	11
41	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. <i>Brain</i> , 2019, 142, 586-605.	7.6	84
42	Cerebellar tract alterations in PLS and ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 281-284.	1.7	26
43	The relationships between symptoms, disability, perceived health and quality of life in amyotrophic lateral sclerosis/motor neuron disease. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 317-327.	1.7	27
44	CSF chitinase proteins in amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 1215-1220.	1.9	66
45	Neuronal over-expression of Oxr1 is protective against ALS-associated mutant TDP-43 mislocalisation in motor neurons and neuromuscular defects in vivo. <i>Human Molecular Genetics</i> , 2019, 28, 3584-3599.	2.9	19
46	Interaction of Axonal Chondrolectin with Collagen XIXa1 Is Necessary for Precise Neuromuscular Junction Formation. <i>Cell Reports</i> , 2019, 29, 1082-1098.e10.	6.4	13
47	Frequency and signature of somatic variants in 1461 human brain exomes. <i>Genetics in Medicine</i> , 2019, 21, 904-912.	2.4	20
48	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. <i>Neurobiology of Disease</i> , 2019, 121, 148-162.	4.4	62
49	Objectively Monitoring Amyotrophic Lateral Sclerosis Patient Symptoms During Clinical Trials With Sensors: Observational Study. <i>JMIR MHealth and UHealth</i> , 2019, 7, e13433.	3.7	32
50	Towards a TDP-43-Based Biomarker for ALS and FTL. <i>Molecular Neurobiology</i> , 2018, 55, 7789-7801.	4.0	100
51	Development and validation of Spasticity Index-Amyotrophic Lateral Sclerosis. <i>Acta Neurologica Scandinavica</i> , 2018, 138, 47-54.	2.1	7
52	Increased cerebral functional connectivity in ALS. <i>Neurology</i> , 2018, 90, e1418-e1424.	1.1	26
53	Impaired corticomuscular and interhemispheric cortical beta oscillation coupling in amyotrophic lateral sclerosis. <i>Clinical Neurophysiology</i> , 2018, 129, 1479-1489.	1.5	36
54	Advances in therapy for spinal muscular atrophy: promises and challenges. <i>Nature Reviews Neurology</i> , 2018, 14, 214-224.	10.1	174

#	ARTICLE	IF	CITATIONS
55	HDAC6 is a therapeutic target in mutant GARS-induced Charcot-Marie-Tooth disease. <i>Brain</i> , 2018, 141, 673-687.	7.6	93
56	The two-year progression of structural and functional cerebral MRI in amyotrophic lateral sclerosis. <i>NeuroImage: Clinical</i> , 2018, 17, 953-961.	2.7	100
57	Cerebrospinal fluid macrophage biomarkers in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2018, 83, 258-268.	5.3	107
58	Oligogenic genetic variation of neurodegenerative disease genes in 980 postmortem human brains. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 813-816.	1.9	17
59	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. <i>Lancet Neurology</i> , The, 2018, 17, 423-433.	10.2	342
60	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
61	UFLCâ€Derived CSF Extracellular Vesicle Origin and Proteome. <i>Proteomics</i> , 2018, 18, e1800257.	2.2	36
62	Neurodegeneration in SCA14 is associated with increased PKCÎ³ kinase activity, mislocalization and aggregation. <i>Acta Neuropathologica Communications</i> , 2018, 6, 99.	5.2	37
63	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.	3.1	59
64	Initial Identification of a Blood-Based Chromosome Conformation Signature for Aiding in the Diagnosis of Amyotrophic Lateral Sclerosis. <i>EBioMedicine</i> , 2018, 33, 169-184.	6.1	17
65	Regional thalamic MRI as a marker of widespread cortical pathology and progressive frontotemporal involvement in amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1250-1258.	1.9	39
66	Amyotrophic lateral sclerosis: the complex path to precision medicine. <i>Journal of Neurology</i> , 2018, 265, 2454-2462.	3.6	36
67	Sweet food preference in amyotrophic lateral sclerosis. <i>Practical Neurology</i> , 2017, 17, 128-129.	1.1	4
68	A risk stratifying tool to facilitate safe late-stage percutaneous endoscopic gastrostomy in ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 243-248.	1.7	18
69	ADCY5-related dyskinesia presenting as familial myoclonus-dystonia. <i>Neurogenetics</i> , 2017, 18, 111-117.	1.4	31
70	Mitochondrial DNA point mutations and relative copy number in 1363 disease and control human brains. <i>Acta Neuropathologica Communications</i> , 2017, 5, 13.	5.2	83
71	Whole-exome sequencing of 228 patients with sporadic Parkinsonâ€™s disease. <i>Scientific Reports</i> , 2017, 7, 41188.	3.3	27
72	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, .	12.4	129

#	ARTICLE	IF	CITATIONS
73	The SMA Trust: the role of a disease-focused research charity in developing treatments for SMA. <i>Gene Therapy</i> , 2017, 24, 544-546.	4.5	8
74	C9orf72 and RAB7L1 regulate vesicle trafficking in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Brain</i> , 2017, 140, 887-897.	7.6	126
75	Trk receptor signaling and sensory neuron fate are perturbed in human neuropathy caused by <i>Gars</i> mutations. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, E3324-E3333.	7.1	61
76	Genetic compendium of 1511 human brains available through the UK Medical Research Council Brain Banks Network Resource. <i>Genome Research</i> , 2017, 27, 165-173.	5.5	44
77	July 2017 ENCALS statement on edaravone. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 471-474.	1.7	41
78	Therapeutic strategies for spinal muscular atrophy: SMN and beyond. <i>DMM Disease Models and Mechanisms</i> , 2017, 10, 943-954.	2.4	87
79	Fundus fluorescein angiography in Susac's syndrome. <i>Practical Neurology</i> , 2017, 17, 472-473.	1.1	1
80	Spinal muscular atrophy: antisense oligonucleotide therapy opens the door to an integrated therapeutic landscape. <i>Human Molecular Genetics</i> , 2017, 26, R151-R159.	2.9	61
81	The clinical landscape for SMA in a new therapeutic era. <i>Gene Therapy</i> , 2017, 24, 529-533.	4.5	133
82	Genetic screening in sporadic ALS and FTD. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 1042-1044.	1.9	105
83	Defective cholesterol metabolism in amyotrophic lateral sclerosis. <i>Journal of Lipid Research</i> , 2017, 58, 267-278.	4.2	115
84	A multicentre evaluation of oropharyngeal secretion management practices in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 1-9.	1.7	20
85	Altered cortical beta-band oscillations reflect motor system degeneration in amyotrophic lateral sclerosis. <i>Human Brain Mapping</i> , 2017, 38, 237-254.	3.6	58
86	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017, 140, 3191-3203.	7.6	323
87	Quantitative FLAIR MRI in Amyotrophic Lateral Sclerosis. <i>Academic Radiology</i> , 2017, 24, 1187-1194.	2.5	31
88	ABN news. <i>Practical Neurology</i> , 2016, 16, 171-171.	1.1	0
89	Vascular effects and spinal cord hypoxia in spinal muscular atrophy. <i>Annals of Neurology</i> , 2016, 79, 217-230.	5.3	79
90	Clinical tool for predicting survival in ALS: do we need one?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 1275-1275.	1.9	4

#	ARTICLE	IF	CITATIONS
91	What is the role of TDP-43 in <i>C9orf72</i> -related amyotrophic lateral sclerosis and frontotemporal dementia?. <i>Brain</i> , 2016, 139, 3057-3059.	7.6	7
92	Increased functional connectivity common to symptomatic amyotrophic lateral sclerosis and those at genetic risk. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 580-588.	1.9	82
93	Extracellular vesicles in neurodegenerative disease – pathogenesis to biomarkers. <i>Nature Reviews Neurology</i> , 2016, 12, 346-357.	10.1	299
94	Pathogenesis of FUS-associated ALS and FTD: insights from rodent models. <i>Acta Neuropathologica Communications</i> , 2016, 4, 99.	5.2	97
95	<i>C9orf72</i> 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. <i>Stem Cells</i> , 2016, 34, 2063-2078.	3.2	195
96	Systemic peptide-mediated oligonucleotide therapy improves long-term survival in spinal muscular atrophy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 10962-10967.	7.1	159
97	Nutritional pathway for people with motor neurone disease. <i>British Journal of Community Nursing</i> , 2016, 21, 360-363.	0.4	1
98	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	21.4	218
99	Psychiatric disorders prior to amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2016, 80, 935-938.	5.3	49
100	Cerebrovascular injury as a risk factor for amyotrophic lateral sclerosis: Table 1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 244-246.	1.9	31
101	The ALSFRS as an outcome measure in therapeutic trials and its relationship to symptom onset. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2016, 17, 414-425.	1.7	65
102	ER Stress and Autophagic Perturbations Lead to Elevated Extracellular α -Synuclein in GBA-N370S Parkinson's iPSC-Derived Dopamine Neurons. <i>Stem Cell Reports</i> , 2016, 6, 342-356.	4.8	279
103	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	6.2	333
104	Eye-tracking in amyotrophic lateral sclerosis: A longitudinal study of saccadic and cognitive tasks. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2016, 17, 101-111.	1.7	65
105	DiPALS: Diaphragm Pacing in patients with Amyotrophic Lateral Sclerosis – a randomised controlled trial. <i>Health Technology Assessment</i> , 2016, 20, 1-186.	2.8	13
106	Identification of distinct circulating exosomes in Parkinson's disease. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 353-361.	3.7	111
107	CSF neurofilament light chain reflects corticospinal tract degeneration in ALS. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 748-755.	3.7	118
108	Motor neuron disease: current management and future prospects. <i>Internal Medicine Journal</i> , 2015, 45, 1005-1013.	0.8	18

#	ARTICLE	IF	CITATIONS
109	Improving access to medicines: empowering patients in the quest to improve treatment for rare lethal diseases. <i>Journal of Medical Ethics</i> , 2015, 41, 987-989.	1.8	8
110	TARDBP pathogenic mutations increase cytoplasmic translocation of TDP-43 and cause reduction of endoplasmic reticulum Ca ²⁺ signaling in motor neurons. <i>Neurobiology of Disease</i> , 2015, 75, 64-77.	4.4	45
111	The longitudinal cerebrospinal fluid metabolomic profile of amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2015, 16, 456-463.	1.7	49
112	Dominant, toxic gain-of-function mutations in <i>gars</i> lead to non-cell autonomous neuropathology. <i>Human Molecular Genetics</i> , 2015, 24, 4397-4406.	2.9	50
113	Neurofilament light chain. <i>Neurology</i> , 2015, 84, 2247-2257.	1.1	412
114	Safety and efficacy of diaphragm pacing in patients with respiratory insufficiency due to amyotrophic lateral sclerosis (DiPALS): a multicentre, open-label, randomised controlled trial. <i>Lancet Neurology</i> , The, 2015, 14, 883-892.	10.2	85
115	² -Weighted MRI Detects Presymptomatic Pathology in the SOD1 Mouse Model of ALS. <i>Journal of Cerebral Blood Flow and Metabolism</i> , 2014, 34, 785-793.	4.3	32
116	Expanding the genetics of huntingtonism. <i>Neurology</i> , 2014, 82, 286-287.	1.1	5
117	Widespread grey matter pathology dominates the longitudinal cerebral MRI and clinical landscape of amyotrophic lateral sclerosis. <i>Brain</i> , 2014, 137, 2546-2555.	7.6	151
118	An impedimetric assay of α -synuclein autoantibodies in early stage Parkinson's disease. <i>RSC Advances</i> , 2014, 4, 58773-58777.	3.6	18
119	Predictors of cognitive impairment in an early stage Parkinson's disease cohort. <i>Movement Disorders</i> , 2014, 29, 351-359.	3.9	124
120	Advances in the clinical science of the motor unit. <i>Current Opinion in Neurology</i> , 2014, 27, 503-505.	3.6	0
121	Swallowing and oropharyngeal dysphagia. <i>Clinical Medicine</i> , 2014, 14, 456.	1.9	2
122	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.	1.7	15
123	Comprehensive morphometry of subcortical grey matter structures in early-stage Parkinson's disease. <i>Human Brain Mapping</i> , 2014, 35, 1681-1690.	3.6	84
124	Chondrolectin affects cell survival and neuronal outgrowth in in vitro and in vivo models of spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2014, 23, 855-869.	2.9	62
125	Advances in motor neurone disease. <i>Journal of the Royal Society of Medicine</i> , 2014, 107, 14-21.	2.0	93
126	Neuromuscular junction maturation defects precede impaired lower motor neuron connectivity in Charcot-Marie-Tooth type 2D mice. <i>Human Molecular Genetics</i> , 2014, 23, 2639-2650.	2.9	75

#	ARTICLE	IF	CITATIONS
127	A blinded international study on the reliability of genetic testing for GGGCC-repeat expansions in <i>C9orf72</i> reveals marked differences in results among 14 laboratories. <i>Journal of Medical Genetics</i> , 2014, 51, 419-424.	3.2	118
128	REM sleep behaviour disorder is associated with worse quality of life and other non-motor features in early Parkinson's disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 560-566.	1.9	150
129	Overexpression of survival motor neuron improves neuromuscular function and motor neuron survival in mutant SOD1 mice. <i>Neurobiology of Aging</i> , 2014, 35, 906-915.	3.1	39
130	Progressive hemiparesis (Mills syndrome) with aphasia in amyotrophic lateral sclerosis. <i>Neurology</i> , 2014, 82, 457-458.	1.1	15
131	Amyotrophic lateral sclerosis: cell vulnerability or system vulnerability?. <i>Journal of Anatomy</i> , 2014, 224, 45-51.	1.5	32
132	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	8.1	308
133	FTLD-ALS of TDP-43 type and SCA2 in a family with a full ataxin-2 polyglutamine expansion. <i>Acta Neuropathologica</i> , 2014, 128, 597-604.	7.7	23
134	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. <i>Parkinsonism and Related Disorders</i> , 2014, 20, 99-105.	2.2	223
135	Should all patients with ALS have genetic testing?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 475-475.	1.9	5
136	SMA-EUROPE workshop report: opportunities and challenges in developing clinical trials for spinal muscular atrophy in Europe. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 44.	2.7	17
137	Unmasking of incipient amyotrophic lateral sclerosis by botulinum toxin therapy. <i>Journal of Neurology</i> , 2013, 260, 1166-1167.	3.6	7
138	Progressive ataxia and palatal tremor associated with dense pontine calcification: A unique case. <i>Movement Disorders</i> , 2013, 28, 1155-1157.	3.9	5
139	Assessing social isolation in motor neurone disease: A Rasch analysis of the MND Social Withdrawal Scale. <i>Journal of the Neurological Sciences</i> , 2013, 334, 112-118.	0.6	8
140	Management of sialorrhoea in motor neuron disease: A survey of current UK practice. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 521-527.	1.7	28
141	Controversies and priorities in amyotrophic lateral sclerosis. <i>Lancet Neurology</i> , The, 2013, 12, 310-322.	10.2	454
142	A Loss-of-Function Variant in the Human Histidyl-tRNA Synthetase (<i>HARS</i>) Gene is Neurotoxic In Vivo. <i>Human Mutation</i> , 2013, 34, 191-199.	2.5	104
143	Lithium in patients with amyotrophic lateral sclerosis (LiCALS): a phase 3 multicentre, randomised, double-blind, placebo-controlled trial. <i>Lancet Neurology</i> , The, 2013, 12, 339-345.	10.2	118
144	Mimics and chameleons in motor neurone disease. <i>Practical Neurology</i> , 2013, 13, 153-164.	1.1	84

#	ARTICLE	IF	CITATIONS
145	Next generation sequencing for molecular diagnosis of neurological disorders using ataxias as a model. <i>Brain</i> , 2013, 136, 3106-3118.	7.6	146
146	Whole-brain magnetic resonance spectroscopic imaging measures are related to disability in ALS. <i>Neurology</i> , 2013, 80, 610-615.	1.1	50
147	Myelin imaging in amyotrophic and primary lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 562-573.	1.7	59
148	Autoimmune disease preceding amyotrophic lateral sclerosis. <i>Neurology</i> , 2013, 81, 1222-1225.	1.1	156
149	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, 666-673.	1.9	43
150	The impact of fatigue and psychosocial variables on quality of life for patients with motor neuron disease. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 537-545.	1.7	31
151	An Eye-Tracking Version of the Trail-Making Test. <i>PLoS ONE</i> , 2013, 8, e84061.	2.5	21
152	Cardiovascular fitness as a risk factor for amyotrophic lateral sclerosis: indirect evidence from record linkage study: Table 1. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2012, 83, 395-398.	1.9	62
153	Fractional Anisotropy in the Posterior Limb of the Internal Capsule and Prognosis in Amyotrophic Lateral Sclerosis. <i>Archives of Neurology</i> , 2012, 69, 1493.	4.5	60
154	Motor neurone disease is a clinical diagnosis. <i>Practical Neurology</i> , 2012, 12, 396-397.	1.1	6
155	Systematic review of methodology used in ultrasound studies aimed at creating charts of fetal size. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2012, 119, 1425-1439.	2.3	119
156	Magnetic resonance imaging of pathological processes in rodent models of amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2012, 13, 288-301.	2.1	8
157	Protocol for diaphragm pacing in patients with respiratory muscle weakness due to motor neurone disease (DiPALS): a randomised controlled trial. <i>BMC Neurology</i> , 2012, 12, 74.	1.8	7
158	A calm before the exome storm. <i>Neurology</i> , 2012, 78, 1706-1707.	1.1	6
159	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. <i>Lancet Neurology</i> , The, 2012, 11, 323-330.	10.2	1,039
160	A Recurrent loss-of-function alanyl-tRNA synthetase (AARS) mutation in patients with charcot-marie-tooth disease type 2N (CMT2N). <i>Human Mutation</i> , 2012, 33, 244-253.	2.5	90
161	Heterotopic pregnancy. <i>Journal of Obstetrics and Gynaecology</i> , 2011, 31, 7-12.	0.9	109
162	HspB8 mutation causing hereditary distal motor neuropathy impairs lysosomal delivery of autophagosomes. <i>Journal of Neurochemistry</i> , 2011, 119, 1155-1161.	3.9	49

#	ARTICLE	IF	CITATIONS
163	Development of a patient reported outcome measure for fatigue in motor neurone disease: the Neurological Fatigue Index (NFI-MND). <i>Health and Quality of Life Outcomes</i> , 2011, 9, 101.	2.4	31
164	Rasch analysis of the hospital anxiety and depression scale (hads) for use in motor neurone disease. <i>Health and Quality of Life Outcomes</i> , 2011, 9, 82.	2.4	96
165	Integration of structural and functional magnetic resonance imaging in amyotrophic lateral sclerosis. <i>Brain</i> , 2011, 134, 3470-3479.	7.6	229
166	Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2011, 20, 4334-4344.	2.9	89
167	Concordance between site of onset and limb dominance in amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 853-854.	1.9	114
168	Oculomotor Dysfunction in Amyotrophic Lateral Sclerosis. <i>Archives of Neurology</i> , 2011, 68, 857.	4.5	112
169	The contribution of mouse models to understanding the pathogenesis of spinal muscular atrophy. <i>DMM Disease Models and Mechanisms</i> , 2011, 4, 457-467.	2.4	113
170	Familial versus sporadic amyotrophic lateral sclerosis--a false dichotomy?. <i>Brain</i> , 2011, 134, 3429-3434.	7.6	22
171	Catastrophic hyperkalaemia following administration of suxamethonium chloride to a patient with undiagnosed amyotrophic lateral sclerosis. <i>Clinical Medicine</i> , 2011, 11, 292-293.	1.9	10
172	Charcot-Marie-Toothâ€“Linked Mutant GARS Is Toxic to Peripheral Neurons Independent of Wild-Type GARS Levels. <i>PLoS Genetics</i> , 2011, 7, e1002399.	3.5	109
173	Compound Heterozygosity for Loss-of-Function Lysyl-tRNA Synthetase Mutations in a Patient with Peripheral Neuropathy. <i>American Journal of Human Genetics</i> , 2010, 87, 560-566.	6.2	169
174	Novel SPG11 mutations in Asian kindreds and disruption of spatascin function in the zebrafish. <i>Neurogenetics</i> , 2010, 11, 379-389.	1.4	26
175	Review: Neuromuscular synaptic vulnerability in motor neurone disease: amyotrophic lateral sclerosis and spinal muscular atrophy. <i>Neuropathology and Applied Neurobiology</i> , 2010, 36, 133-156.	3.2	123
176	Robustness Testing against Low Voltage Transients - A Novel Approach. , 2010, , .		3
177	PATU5 Characterisation of fused in sarcoma pathology and FUS mutations in juvenile amyotrophic lateral sclerosis with basophilic inclusions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, e25-e25.	1.9	0
178	Pre-symptomatic development of lower motor neuron connectivity in a mouse model of severe spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2010, 19, 420-433.	2.9	98
179	SMN deficiency disrupts brain development in a mouse model of severe spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2010, 19, 4216-4228.	2.9	105
180	Dismutase-competent SOD1 mutant accumulation in myelinating Schwann cells is not detrimental to normal or transgenic ALS model mice. <i>Human Molecular Genetics</i> , 2010, 19, 815-824.	2.9	52

#	ARTICLE	IF	CITATIONS
181	Reduction of elevated IGF-1 levels in coincident amyotrophic lateral sclerosis and acromegaly. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 255-257.	2.1	1
182	Corpus callosum involvement is a consistent feature of amyotrophic lateral sclerosis. <i>Neurology</i> , 2010, 75, 1645-1652.	1.1	257
183	The role of RNA processing in the pathogenesis of motor neuron degeneration. <i>Expert Reviews in Molecular Medicine</i> , 2010, 12, e21.	3.9	29
184	POMD09 Understanding the early pathological pathways in Parkinson's disease. The Oxford Parkinson's Disease Centre. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, e59-e59.	1.9	0
185	Juvenile ALS with basophilic inclusions is a FUS proteinopathy with <i>FUS</i> mutations. <i>Neurology</i> , 2010, 75, 611-618.	1.1	178
186	Do twin studies still have anything to teach us about the genetics of amyotrophic lateral sclerosis?. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 1299-1300.	1.9	3
187	Head and other physical trauma requiring hospitalisation is not a significant risk factor in the development of ALS. <i>Journal of the Neurological Sciences</i> , 2010, 288, 45-48.	0.6	48
188	The diagnostic pathway and prognosis in bulbar-onset amyotrophic lateral sclerosis. <i>Journal of the Neurological Sciences</i> , 2010, 294, 81-85.	0.6	87
189	GARS axonopathy: not every neuron's cup of tRNA. <i>Trends in Neurosciences</i> , 2010, 33, 59-66.	8.6	70
190	Pattern of spread and prognosis in lower limb-onset ALS. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , 2010, 11, 369-373.	2.1	58
191	TARDBP in amyotrophic lateral sclerosis: identification of a novel variant but absence of copy number variation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2009, 80, 1283-1285.	1.9	28
192	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009, 18, 1524-1532.	2.9	106
193	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. <i>DMM Disease Models and Mechanisms</i> , 2009, 2, 359-373.	2.4	91
194	Alternative Splicing Events Are a Late Feature of Pathology in a Mouse Model of Spinal Muscular Atrophy. <i>PLoS Genetics</i> , 2009, 5, e1000773.	3.5	210
195	Another gene for ALS. <i>Neurology</i> , 2009, 73, 1172-1173.	1.1	11
196	Survival motor neuron deficiency enhances progression in an amyotrophic lateral sclerosis mouse model. <i>Neurobiology of Disease</i> , 2009, 34, 511-517.	4.4	62
197	Biomarkers in amyotrophic lateral sclerosis. <i>Lancet Neurology</i> , The, 2009, 8, 94-109.	10.2	391
198	Investigation of white matter pathology in ALS and PLS using tract-based spatial statistics. <i>Human Brain Mapping</i> , 2009, 30, 615-624.	3.6	123

#	ARTICLE	IF	CITATIONS
199	Motor neuron disease: THE BARE ESSENTIALS. <i>Practical Neurology</i> , 2009, 9, 303-309.	1.1	115
200	Functional vitamin B12 deficiency. <i>Practical Neurology</i> , 2009, 9, 37-45.	1.1	33
201	Progressive unsteadiness in a 68-year-old man with longstanding abdominal pain and altered bowel habit. <i>Practical Neurology</i> , 2009, 9, 210-220.	1.1	4
202	Primary angiitis of the CNS mimicking a spinal cord tumour. <i>Journal of Neurology</i> , 2008, 255, 1970-1972.	3.6	4
203	Asymmetrical late onset motor neuropathy associated with a novel mutation in the small heat shock protein HSPB1 (HSP27). <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2008, 79, 461-463.	1.9	41
204	TDP-43 expression in mouse models of amyotrophic lateral sclerosis and spinal muscular atrophy. <i>BMC Neuroscience</i> , 2008, 9, 104.	1.9	55
205	Candidate screening of the bovine and feline spinal muscular atrophy genes reveals no evidence for involvement in human motor neuron disorders. <i>Neuromuscular Disorders</i> , 2008, 18, 394-397.	0.6	7
206	Transgenics, toxicity and therapeutics in rodent models of mutant SOD1-mediated familial ALS. <i>Progress in Neurobiology</i> , 2008, 85, 94-134.	5.7	435
207	Activation of mutant protein kinase C δ^3 leads to aberrant sequestration and impairment of its cellular function. <i>Biochemical and Biophysical Research Communications</i> , 2008, 372, 447-453.	2.1	9
208	Is Good Housekeeping the Key to Motor Neuron Survival?. <i>Cell</i> , 2008, 133, 572-574.	28.9	14
209	Selective vulnerability of motor neurons and dissociation of pre- and post-synaptic pathology at the neuromuscular junction in mouse models of spinal muscular atrophy. <i>Human Molecular Genetics</i> , 2008, 17, 949-962.	2.9	333
210	A case of celiac disease mimicking amyotrophic lateral sclerosis. <i>Nature Clinical Practice Neurology</i> , 2007, 3, 581-584.	2.5	26
211	Magnetic resonance spectroscopic imaging of prognostic value in amyotrophic lateral sclerosis?. <i>Nature Clinical Practice Neurology</i> , 2007, 3, 76-77.	2.5	1
212	Genetics of sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2007, 16, R233-R242.	2.9	145
213	Chapter 7 Spinal muscular atrophies and hereditary motor neuropathies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2007, 82, 141-153.	1.8	3
214	The study of rare diseases: butterfly collecting or an entree to understanding common conditions?. <i>Practical Neurology</i> , 2007, 7, 210-211.	1.1	3
215	Crystal structure of human wildtype and S581L-mutant glycyl-tRNA synthetase, an enzyme underlying distal spinal muscular atrophy. <i>FEBS Letters</i> , 2007, 581, 2959-2964.	2.8	28
216	Murray Valley encephalitis in an adult traveller complicated by long-term flaccid paralysis: case report and review of the literature. <i>Transactions of the Royal Society of Tropical Medicine and Hygiene</i> , 2007, 101, 284-288.	1.8	19

#	ARTICLE	IF	CITATIONS
217	Neuromuscular disorders: therapeutic advances. <i>Lancet Neurology</i> , The, 2007, 6, 18-19.	10.2	1
218	The molecular genetics of non-ALS motor neuron diseases. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2006, 1762, 986-1000.	3.8	41
219	Treatment of vasculitic peripheral neuropathy: a retrospective analysis of outcome. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2006, 100, 41-51.	0.5	59
220	Recent advances in the genetics of amyotrophic lateral sclerosis and frontotemporal dementia: common pathways in neurodegenerative disease. <i>Human Molecular Genetics</i> , 2006, 15, R182-R187.	2.9	81
221	A mutation in the small heat-shock protein HSPB1 leading to distal hereditary motor neuropathy disrupts neurofilament assembly and the axonal transport of specific cellular cargoes. <i>Human Molecular Genetics</i> , 2006, 15, 347-354.	2.9	138
222	Severe childhood SMA and axonal CMT due to anticodon binding domain mutations in the GARS gene. <i>Neurology</i> , 2006, 67, 1710-1712.	1.1	72
223	Hot-spot residue in small heat-shock protein 22 causes distal motor neuropathy. <i>Nature Genetics</i> , 2004, 36, 597-601.	21.4	395
224	Mutant small heat-shock protein 27 causes axonal Charcot-Marie-Tooth disease and distal hereditary motor neuropathy. <i>Nature Genetics</i> , 2004, 36, 602-606.	21.4	541
225	Vitamin E deficiency induced neurological disease in common variable immunodeficiency: two cases and a review of the literature of vitamin E deficiency. <i>Clinical Immunology</i> , 2004, 112, 24-29.	3.2	67
226	Motor neuron disease. <i>Medicine</i> , 2004, 32, 105-107.	0.4	2
227	Musculoskeletal diseases: from complex genetics to therapy. <i>Current Opinion in Pharmacology</i> , 2003, 3, 277-279.	3.5	0
228	Reduction in excess daytime sleepiness by modafinil in patients with myotonic dystrophy. <i>Neuromuscular Disorders</i> , 2003, 13, 357-364.	0.6	83
229	Spinal muscular atrophies reveal motor neuron vulnerability to defects in ribonucleoprotein handling. <i>Current Opinion in Neurology</i> , 2003, 16, 595-599.	3.6	17
230	Chapter 16 Spinal Muscular Atrophy. <i>Blue Books of Practical Neurology</i> , 2003, 28, 401-cp2.	0.1	1
231	Motor neurone disease. <i>Postgraduate Medical Journal</i> , 2002, 78, 513-519.	1.8	92
232	Normal d-dimer levels do not exclude the diagnosis of cerebral venous sinus thrombosis. <i>Journal of Neurology</i> , 2002, 249, 1603-1604.	3.6	27
233	Spinal Muscular Atrophy. <i>Seminars in Neurology</i> , 2001, 21, 189-198.	1.4	68
234	Screwdriver Headache: A Case of Traumatic Intracranial Hypotension. <i>Clinical Radiology</i> , 2001, 56, 676-680.	1.1	13

#	ARTICLE	IF	CITATIONS
235	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
236	Spinal muscular atrophy. Journal of Inherited Metabolic Disease, 1999, 22, 545-554.	3.6	33
237	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
238	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
239	Genomic Organization and Chromosomal Localization of a Member of the MAP Kinase Phosphatase Gene Family to Human Chromosome 11p15.5 and a Pseudogene to 10q11.2. Genomics, 1997, 42, 284-294.	2.9	15
240	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
241	What's new in the molecular genetics of spinal muscular atrophy?. European Journal of Paediatric Neurology, 1997, 1, 149-155.	1.6	2
242	Gene deletions in spinal muscular atrophy.. Journal of Medical Genetics, 1996, 33, 93-96.	3.2	77
243	Molecular Genetics of Autosomal Recessive Spinal Muscular Atrophy. Molecular Medicine, 1996, 2, 400-404.	4.4	3
244	Evidence for compound heterozygosity causing mild and severe forms of autosomal recessive spinal muscular atrophy.. Journal of Medical Genetics, 1996, 33, 1019-1021.	3.2	4
245	Molecular genetics of autosomal recessive spinal muscular atrophy. Molecular Medicine, 1996, 2, 400-4.	4.4	1
246	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
247	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
248	Motor disorders. , 0, , 63-78.		0
249	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
250	ALS Mice Carrying Pathological Mutant TDP-43, But Not Mutant FUS, Display Axonal Transport Defects <i>in vivo</i>. SSRN Electronic Journal, 0, , .	0.4	1