Pamela Shaw

List of Publications by Year in Descending Order

406 papers	25,389 citations	87 h-index	143 g-index
450 ext. papers	29,950 ext. citations	7.3 avg, IF	6.81 L-index

#	Paper	IF	Citations
406	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis.. *Neuron*, **2022**,	13.9	8
405	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis.. *Npj Genomic Medicine*, **2022**, 7, 8	6.2	4
404	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain.. *Brain Communications*, **2022**, 4, fcac029	4.5	2
403	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS.. *Science Translational Medicine*, **2022**, 14, eabj0264	17.5	4
402	Unbiased metabolome screen leads to personalized medicine strategy for amyotrophic lateral sclerosis.. *Brain Communications*, **2022**, 4, fcac069	4.5	1
401	Tensor electrical impedance myography identifies bulbardisease progression in amyotrophic lateral sclerosis.. *Clinical Neurophysiology*, **2022**, 139, 69-75	4.3	
400	Concurrent sodium channel myotonia and amyotrophic lateral sclerosis supports shared pathogenesis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2022**, 93, A13.2-A13	5.5	
399	Simultaneous ALS and SCA2 associated with an intermediate-length CAG-repeat expansion. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2021**, 22, 579-582	3.6	3
398	Common Genetic Variants Contribute to Risk of Transposition of the Great Arteries. *Circulation Research*, **2021**,	15.7	2
397	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. *Nature Genetics*, **2021**, 53, 1636-1648	36.3	19
396	Proteinopathies as Hallmarks of Impaired Gene Expression, Proteostasis and Mitochondrial Function in Amyotrophic Lateral Sclerosis.. *Frontiers in Neuroscience*, **2021**, 15, 783624	5.1	1
395	A review of Mendelian randomization in amyotrophic lateral sclerosis. *Brain*, **2021**,	11.2	7
394	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. *Genome Biology*, **2021**, 22, 90	18.3	6
393	Fiber Optic Raman Spectroscopy of Muscle in Preclinical Models of Amyotrophic Lateral Sclerosis and Duchenne Muscular Dystrophy. *ACS Chemical Neuroscience*, **2021**, 12, 1768-1776	5.7	2
392	Adipose-derived stem cells protect motor neurons and reduce glial activation in both and models of ALS. *Molecular Therapy - Methods and Clinical Development*, **2021**, 21, 413-433	6.4	3
391	Proteomic Approaches to Study Cysteine Oxidation: Applications in Neurodegenerative Diseases. *Frontiers in Molecular Neuroscience*, **2021**, 14, 678837	6.1	3
390	Physical exercise is a risk factor for amyotrophic lateral sclerosis: Convergent evidence from Mendelian randomisation, transcriptomics and risk genotypes. *EBioMedicine*, **2021**, 68, 103397	8.8	19

389	Directly converted astrocytes retain the ageing features of the donor fibroblasts and elucidate the astrocytic contribution to human CNS health and disease. *Aging Cell*, **2021**, 20, e13281	9.9	10
388	Fit for purpose? A cross-sectional study to evaluate the acceptability and usability of HeadUp, a novel neck support collar for neurological neck weakness. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2021**, 22, 38-45	3.6	
387	The Effect of SMN Gene Dosage on ALS Risk and Disease Severity. *Annals of Neurology*, **2021**, 89, 686-697	9.4	2
386	The gut microbiome: a key player in the complexity of amyotrophic lateral sclerosis (ALS). *BMC Medicine*, **2021**, 19, 13	11.4	16
385	Mitochondrial Dysfunction in Alzheimer's Disease: A Biomarker of the Future?. *Biomedicines*, **2021**, 9,	4.8	19
384	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**, 92, 510-518	5.5	20
383	Innovating Clinical Trials for Amyotrophic Lateral Sclerosis: Challenging the Established Order. *Neurology*, **2021**, 97, 528-536	6.5	2
382	SRSF1-dependent inhibition of C9ORF72-repeat RNA nuclear export: genome-wide mechanisms for neuroprotection in amyotrophic lateral sclerosis. *Molecular Neurodegeneration*, **2021**, 16, 53	19	2
381	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2021**, 78, 1236-1248	17.2	5
380	Extensive phenotypic characterisation of a human TDP-43 transgenic mouse model of amyotrophic lateral sclerosis (ALS). *Scientific Reports*, **2021**, 11, 16659	4.9	0
379	Tensor electrical impedance myography identifies clinically relevant features in amyotrophic lateral sclerosis. *Physiological Measurement*, **2021**, 42,	2.9	2
378	Amyotrophic lateral sclerosis alters the metabolic aging profile in patient derived fibroblasts. *Neurobiology of Aging*, **2021**, 105, 64-77	5.6	4
377	Amyotrophic lateral sclerosis transcriptomics reveals immunological effects of low-dose interleukin-2. *Brain Communications*, **2021**, 3, fcab141	4.5	1
376	Modelling and analysis of electrical impedance myography of the lateral tongue. *Physiological Measurement*, **2021**, 41, 125008	2.9	3
375	Type 2 diabetes mellitus-associated transcriptome alterations in cortical neurones and associated neurovascular unit cells in the ageing brain. *Acta Neuropathologica Communications*, **2021**, 9, 5	7.3	5
374	Do deficits in mitochondrial spare respiratory capacity contribute to neuropsychological changes seen in Alzheimer disease?. *Alzheimerjs and Dementia*, **2020**, 16, e037527	1.2	
373	Deficits in Mitochondrial Spare Respiratory Capacity Contribute to the Neuropsychological Changes of Alzheimer's Disease. *Journal of Personalized Medicine*, **2020**, 10,	3.6	10
372	Oligodendrocyte pathology exceeds axonal pathology in white matter in human amyotrophic lateral sclerosis. *Journal of Pathology*, **2020**, 251, 262-271	9.4	15

371	Phase 1-2 Trial of Antisense Oligonucleotide Tofersen for ALS. *New England Journal of Medicine*, **2020**, 383, 109-119	59.2	140
370	Multi-dimensional electrical impedance myography of the tongue as a potential biomarker for amyotrophic lateral sclerosis. *Clinical Neurophysiology*, **2020**, 131, 799-808	4.3	11
369	Concurrent sodium channelopathies and amyotrophic lateral sclerosis supports shared pathogenesis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2020**, 21, 627-630	3.6	1
368	Magnetic resonance spectroscopy reveals mitochondrial dysfunction in amyotrophic lateral sclerosis. *Brain*, **2020**, 143, 3603-3618	11.2	6
367	repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. *Brain Communications*, **2020**, 2, fcaa064	4.5	12
366	Cross-reactive probes on Illumina DNA methylation arrays: a large study on ALS shows that a cautionary approach is warranted in interpreting epigenome-wide association studies. *NAR Genomics and Bioinformatics*, **2020**, 2, lqaa105	3.7	3
365	Neuropathological characterization of a novel TANK binding kinase (TBK1) gene loss of function mutation associated with amyotrophic lateral sclerosis. *Neuropathology and Applied Neurobiology*, **2020**, 46, 279-291	5.2	6
364	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. *Cell Reports*, **2020**, 33, 108456	10.6	6
363	Advanced Glycation End Product Formation in Human Cerebral Cortex Increases With Alzheimer-Type Neuropathologic Changes but Is Not Independently Associated With Dementia in a Population-Derived Aging Brain Cohort. *Journal of Neuropathology and Experimental Neurology*, **2020**, 79, 950-958	3.1	5
362	Repeated 5-day cycles of low dose aldesleukin in amyotrophic lateral sclerosis (IMODALS): A phase 2a randomised, double-blind, placebo-controlled trial. *EBioMedicine*, **2020**, 59, 102844	8.8	12
361	SOD1-targeting therapies for neurodegenerative diseases: a review of current findings and future potential. *Expert Opinion on Orphan Drugs*, **2020**, 8, 379-392	1.1	0
360	Biomarkers in amyotrophic lateral sclerosis: a review of new developments. *Current Opinion in Neurology*, **2020**, 33, 662-668	7.1	8
359	Longitudinal multi-modal muscle-based biomarker assessment in motor neuron disease. *Journal of Neurology*, **2020**, 267, 244-256	5.5	8
358	UK case control study of smoking and risk of amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2020**, 21, 222-227	3.6	5
357	Health care professionals' views on psychological factors affecting nutritional behaviour in people with motor neuron disease: A thematic analysis. *British Journal of Health Psychology*, **2019**, 24, 953-969	8.3	14
356	Combined fused in sarcoma-positive (FUS+) basophilic inclusion body disease and atypical tauopathy presenting with an amyotrophic lateral sclerosis/motor neurone disease (ALS/MND)-plus phenotype. *Neuropathology and Applied Neurobiology*, **2019**, 45, 586-596	5.2	5
355	Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. *Brain*, **2019**, 142, 586-605	11.2	44
354	Telomere length is greater in ALS than in controls: a whole genome sequencing study. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2019**, 20, 229-234	3.6	11

353	Biomarkers in Motor Neuron Disease: A State of the Art Review. *Frontiers in Neurology*, **2019**, 10, 291	4.1	52
352	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. *Annals of Neurology*, **2019**, 85, 470-481	9.4	72
351	Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. *Cell Reports*, **2019**, 26, 2298-2306.e5	10.6	31
350	Differentiation of human adipose-derived stem cells into neuron/motoneuron-like cells for cell replacement therapy of spinal cord injury. *Cell Death and Disease*, **2019**, 10, 597	9.8	36
349	Needs and preferences for psychological interventions of people with motor neuron disease. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2019**, 20, 521-531	3.6	8
348	C9orf72 intermediate expansions of 24-30 repeats are associated with ALS. *Acta Neuropathologica Communications*, **2019**, 7, 115	7.3	35
347	C9orf72 expansion within astrocytes reduces metabolic flexibility in amyotrophic lateral sclerosis. *Brain*, **2019**, 142, 3771-3790	11.2	29
346	Oral levosimendan in amyotrophic lateral sclerosis: a phase II multicentre, randomised, double-blind, placebo-controlled trial. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 1165-1170	5.5	15
345	Using telehealth in motor neuron disease to increase access to specialist multidisciplinary care: a UK-based pilot and feasibility study. *BMJ Open*, **2019**, 9, e028525	3	15
344	Process evaluation and exploration of telehealth in motor neuron disease in a UK specialist centre. *BMJ Open*, **2019**, 9, e028526	3	13
343	Critical design considerations for time-to-event endpoints in amyotrophic lateral sclerosis clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 1331-1337	5.5	6
342	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. *Science Translational Medicine*, **2019**, 11,	17.5	16
341	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. *Nature Neuroscience*, **2019**, 22, 1966-1974	25.5	56
340	Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. *Neurobiology of Aging*, **2019**, 74, 234.e9-234.e15	5.6	13
339	Striking phenotypic variation in a family with the P506S UBQLN2 mutation including amyotrophic lateral sclerosis, spastic paraplegia, and frontotemporal dementia. *Neurobiology of Aging*, **2019**, 73, 229.e5-229.e9	5.6	10
338	Younger age of onset in familial amyotrophic lateral sclerosis is a result of pathogenic gene variants, rather than ascertainment bias. *Journal of Neurology, Neurosurgery and Psychiatry*, **2019**, 90, 268-271	5.5	17
337	The role of mitochondria in amyotrophic lateral sclerosis. *Neuroscience Letters*, **2019**, 710, 132933	3.3	191
336	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. *Lancet Neurology, The*, **2018**, 17, 423-433	24.1	189

335	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
334	The TiM system: developing a novel telehealth service to improve access to specialist care in motor neurone disease using user-centered design. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2018**, 19, 351-361	3.6	29
333	Project MinE: study design and pilot analyses of a large-scale whole-genome sequencing study in amyotrophic lateral sclerosis. *European Journal of Human Genetics*, **2018**, 26, 1537-1546	5.3	75
332	CHCHD10 variants in amyotrophic lateral sclerosis: Where is the evidence?. *Annals of Neurology*, **2018**, 84, 110-116	9.4	16
331	Imaging muscle as a potential biomarker of denervation in motor neuron disease. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 248-255	5.5	25
330	Lost in translation: microRNAs mediate pathological cross-talk between motor neurons and astrocytes. *Brain*, **2018**, 141, 2534-2536	11.2	2
329	Stable transgenic C9orf72 zebrafish model key aspects of the ALS/FTD phenotype and reveal novel pathological features. *Acta Neuropathologica Communications*, **2018**, 6, 125	7.3	28
328	Novel genotype-phenotype and MRI correlations in a large cohort of patients with mutations. *Neurology: Genetics*, **2018**, 4, e279	3.8	32
327	Ursodeoxycholic Acid Improves Mitochondrial Function and Redistributes Drp1 in Fibroblasts from Patients with Either Sporadic or Familial Alzheimer's Disease. *Journal of Molecular Biology*, **2018**, 430, 3942-3953	6.5	35
326	TDP-43 induces p53-mediated cell death of cortical progenitors and immature neurons. *Scientific Reports*, **2018**, 8, 8097	4.9	22
325	Efficacy of the Head Up collar in facilitating functional head movements in patients with Amyotrophic Lateral Sclerosis. *Clinical Biomechanics*, **2018**, 57, 114-120	2.2	5
324	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. *Neurobiology of Aging*, **2018**, 71, 266.e1-266.e10	5.6	44
323	Translating SOD1 Gene Silencing toward the Clinic: A Highly Efficacious, Off-Target-free, and Biomarker-Supported Strategy for fALS. *Molecular Therapy - Nucleic Acids*, **2018**, 12, 75-88	10.7	20
322	C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, 281	5.5	25
321	"Anything that makes life's journey better." Exploring the use of digital technology by people living with motor neurone disease. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2017**, 18, 378-387	3.6	12
320	Comparison of the King's and MiToS staging systems for ALS. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2017**, 18, 227-232	3.6	37
319	Serum miRNAs miR-206, 143-3p and 374b-5p as potential biomarkers for amyotrophic lateral sclerosis (ALS). *Neurobiology of Aging*, **2017**, 55, 123-131	5.6	74
318	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. *Brain*, **2017**, 140, 1611-1618	11.2	46

317	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. *Science Translational Medicine*, **2017**, 9,	17.5	74
316	Viral delivery of hexanucleotide repeat expansions in mice leads to repeat-length-dependent neuropathology and behavioural deficits. *DMM Disease Models and Mechanisms*, **2017**, 10, 859-868	4.1	17
315	A data-driven approach links microglia to pathology and prognosis in amyotrophic lateral sclerosis. *Acta Neuropathologica Communications*, **2017**, 5, 23	7.3	38
314	Amyotrophic lateral sclerosis. *Nature Reviews Disease Primers*, **2017**, 3, 17071	51.1	459
313	July 2017 ENCALS statement on edaravone. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2017**, 18, 471-474	3.6	31
312	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. *Neurology*, **2017**, 89, 1915-1922	6.5	48
311	Advances, challenges and future directions for stem cell therapy in amyotrophic lateral sclerosis. *Molecular Neurodegeneration*, **2017**, 12, 85	19	31
310	Small RNA Sequencing of Sporadic Amyotrophic Lateral Sclerosis Cerebrospinal Fluid Reveals Differentially Expressed miRNAs Related to Neural and Glial Activity. *Frontiers in Neuroscience*, **2017**, 11, 731	5.1	62
309	Can Astrocytes Be a Target for Precision Medicine?. *Advances in Experimental Medicine and Biology*, **2017**, 1007, 111-128	3.6	7
308	Gene Therapy in the Nervous System: Failures and Successes. *Advances in Experimental Medicine and Biology*, **2017**, 1007, 241-257	3.6	2
307	Detection of long repeat expansions from PCR-free whole-genome sequence data. *Genome Research*, **2017**, 27, 1895-1903	9.7	159
306	C9orf72 expansion disrupts ATM-mediated chromosomal break repair. *Nature Neuroscience*, **2017**, 20, 1225-1235	25.5	90
305	SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. *Nature Communications*, **2017**, 8, 16063	17.4	71
304	A multicentre evaluation of oropharyngeal secretion management practices in amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2017**, 18, 1-9	3.6	14
303	PO227 A clinical trial of telehealth to improve timely access to specialist care. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, A72.1-A72	5.5	
302	Protein Homeostasis in Amyotrophic Lateral Sclerosis: Therapeutic Opportunities?. *Frontiers in Molecular Neuroscience*, **2017**, 10, 123	6.1	37
301	Targeted Genetic Screen in Amyotrophic Lateral Sclerosis Reveals Novel Genetic Variants with Synergistic Effect on Clinical Phenotype. *Frontiers in Molecular Neuroscience*, **2017**, 10, 370	6.1	19
300	C9ORF72 hexanucleotide repeat exerts toxicity in a stable, inducible motor neuronal cell model, which is rescued by partial depletion of Pten. *Human Molecular Genetics*, **2017**, 26, 1133-1145	5.6	17

299	An Objective Functional Characterisation of Head Movement Impairment in Individuals with Neck Muscle Weakness Due to Amyotrophic Lateral Sclerosis. *PLoS ONE*, **2017**, 12, e0169019	3.7	6
298	Rare genetic variation in UNC13A may modify survival in amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 593-599	3.6	16
297	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1043-8	36.3	328
296	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1037-42	36.3	149
295	Association of a Locus in the CAMTA1 Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2016**, 73, 812-20	17.2	40
294	Creatine kinase enzyme level correlates positively with serum creatinine and lean body mass, and is a prognostic factor for survival in amyotrophic lateral sclerosis. *European Journal of Neurology*, **2016**, 23, 1071-8	6	31
293	Evidence-based or arrogance-based medicine?. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 305-6	3.6	
292	Long-term physical activity: an exogenous risk factor for sporadic amyotrophic lateral sclerosis?. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 377-84	3.6	31
291	The impact of gastrostomy in motor neurone disease: challenges and benefits from a patient and carer perspective. *BMJ Supportive and Palliative Care*, **2016**, 6, 52-9	2.2	25
290	Forced Expiratory Volume in 1 Second Variability Helps Identify Patients with Cystic Fibrosis at Risk of Greater Loss of Lung Function. *Journal of Pediatrics*, **2016**, 169, 116-21.e2	3.6	33
289	DiPALS: Diaphragm Pacing in patients with Amyotrophic Lateral Sclerosis - a randomised controlled trial. *Health Technology Assessment*, **2016**, 20, 1-186	4.4	5
288	Immune response in peripheral axons delays disease progression in SOD1 mice. *Journal of Neuroinflammation*, **2016**, 13, 261	10.1	44
287	MicroNeurotrophins Improve Survival in Motor Neuron-Astrocyte Co-Cultures but Do Not Improve Disease Phenotypes in a Mutant SOD1 Mouse Model of Amyotrophic Lateral Sclerosis. *PLoS ONE*, **2016**, 11, e0164103	3.7	15
286	Oligogenic inheritance of optineurin (OPTN) and C9ORF72 mutations in ALS highlights localisation of OPTN in the TDP-43-negative inclusions of C9ORF72-ALS. *Neuropathology*, **2016**, 36, 125-34	2	27
285	Motor neurone disease/amyotrophic lateral sclerosis associated with intermediate-length CAG repeat expansions in Ataxin-2 does not have 1C2-positive polyglutamine inclusions. *Neuropathology and Applied Neurobiology*, **2016**, 42, 377-89	5.2	5
284	The role of cranial and thoracic electromyography within diagnostic criteria for amyotrophic lateral sclerosis. *Muscle and Nerve*, **2016**, 54, 378-85	3.4	4
283	The C9orf72 protein interacts with Rab1a and the ULK1 complex to regulate initiation of autophagy. *EMBO Journal*, **2016**, 35, 1656-76	13	237
282	ZNStress: a high-throughput drug screening protocol for identification of compounds modulating neuronal stress in the transgenic mutant sod1G93R zebrafish model of amyotrophic lateral sclerosis. *Molecular Neurodegeneration*, **2016**, 11, 56	19	18

281	Using technology to improve access to specialist care in amyotrophic lateral sclerosis: A systematic review. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 313-24	3.6	29
280	Assessment of the Sheffield Support Snood, an innovative cervical orthosis designed for people affected by neck muscle weakness. *Clinical Biomechanics*, **2016**, 32, 201-6	2.2	14
279	Evaluating a novel cervical orthosis, the Sheffield Support Snood, in patients with amyotrophic lateral sclerosis/motor neuron disease with neck weakness. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 436-42	3.6	16
278	Multicenter validation of CSF neurofilaments as diagnostic biomarkers for ALS. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 404-13	3.6	65
277	Clinical Trials in Neurodegeneration **2016**, 289-303		
276	Case report of concurrent Fabry disease and amyotrophic lateral sclerosis supports a common pathway of pathogenesis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2016**, 17, 614-616	3.6	2
275	Oligodendrocytes contribute to motor neuron death in ALS via SOD1-dependent mechanism. *Proceedings of the National Academy of Sciences of the United States of America*, **2016**, 113, E6496-E6505	11.5	102
274	C9ORF72 interaction with cofilin modulates actin dynamics in motor neurons. *Nature Neuroscience*, **2016**, 19, 1610-1618	25.5	87
273	Clinical aspects of motor neurone disease. *Medicine*, **2016**, 44, 552-556	0.6	11
272	Neuronal DNA damage response-associated dysregulation of signalling pathways and cholesterol metabolism at the earliest stages of Alzheimer-type pathology. *Neuropathology and Applied Neurobiology*, **2016**, 42, 167-79	5.2	21
271	The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. *Neurotherapeutics*, **2015**, 12, 326-39	6.4	39
270	The El Escorial criteria: strengths and weaknesses. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 1-7	3.6	55
269	Developing an outcome measure for excessive saliva management in MND and an evaluation of saliva burden in Sheffield. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 108-13	3.6	6
268	Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 249-51	3.6	6
267	Effect of lipid profile on prognosis in the patients with amyotrophic lateral sclerosis: Insights from the olesoxime clinical trial. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 478-84	3.6	30
266	A preliminary randomized trial of the mechanical insufflator-exsufflator versus breath-stacking technique in patients with amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 448-55	3.6	28
265	Regionality of disease progression predicts prognosis in amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 442-7	3.6	2
264	Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. *Neurobiology of Aging*, **2015**, 36, 2006.e1-9	5.6	14

263	Antisense RNA foci in the motor neurons of C9ORF72-ALS patients are associated with TDP-43 proteinopathy. *Acta Neuropathologica*, **2015**, 130, 63-75	14.3	118
262	Current developments in gene therapy for amyotrophic lateral sclerosis. *Expert Opinion on Biological Therapy*, **2015**, 15, 935-47	5.4	25
261	The role of TREM2 R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. *Alzheimerjs and Dementia*, **2015**, 11, 1407-1416	1.2	126
260	Diagnosis of spinal xanthomatosis by next-generation sequencing: identifying a rare, treatable mimic of hereditary spastic paraparesis. *Practical Neurology*, **2015**, 15, 280-3	2.4	12
259	Cough assistance to clear lungs of ALS patients with severe bulbar dysfunction: Not a good idea!. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2015**, 16, 534-5	3.6	
258	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	24.1	64
257	Altered age-related changes in bioenergetic properties and mitochondrial morphology in fibroblasts from sporadic amyotrophic lateral sclerosis patients. *Neurobiology of Aging*, **2015**, 36, 2893-903	5.6	26
256	The CHCHD10 P34S variant is not associated with ALS in a UK cohort of familial and sporadic patients. *Neurobiology of Aging*, **2015**, 36, 2908.e17-8	5.6	19
255	PTEN depletion decreases disease severity and modestly prolongs survival in a mouse model of spinal muscular atrophy. *Molecular Therapy*, **2015**, 23, 270-7	11.7	40
254	A neuronal DNA damage response is detected at the earliest stages of Alzheimer's neuropathology and correlates with cognitive impairment in the Medical Research Council's Cognitive Function and Ageing Study ageing brain cohort. *Neuropathology and Applied Neurobiology*, **2015**, 41, 483-96	5.2	27
253	Differences in protein quality control correlate with phenotype variability in 2 mouse models of familial amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2015**, 36, 492-504	5.6	43
252	Use of clinical staging in amyotrophic lateral sclerosis for phase 3 clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, **2015**, 86, 45-9	5.5	58
251	Gene expression signatures in motor neurone disease fibroblasts reveal dysregulation of metabolism, hypoxia-response and RNA processing functions. *Neuropathology and Applied Neurobiology*, **2015**, 41, 201-26	5.2	45
250	Invited review: decoding the pathophysiological mechanisms that underlie RNA dysregulation in neurodegenerative disorders: a review of the current state of the art. *Neuropathology and Applied Neurobiology*, **2015**, 41, 109-34	5.2	34
249	LRP-1-mediated intracellular antibody delivery to the Central Nervous System. *Scientific Reports*, **2015**, 5, 11990	4.9	95
248	Lysosomal and phagocytic activity is increased in astrocytes during disease progression in the SOD1 (G93A) mouse model of amyotrophic lateral sclerosis. *Frontiers in Cellular Neuroscience*, **2015**, 9, 410	6.1	25
247	A reduced astrocyte response to β-amyloid plaques in the ageing brain associates with cognitive impairment. *PLoS ONE*, **2015**, 10, e0118463	3.7	36
246	C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. *PLoS ONE*, **2015**, 10, e0127376	3.7	74

245	The nuclear retention of transcription factor FOXO3a correlates with a DNA damage response and increased glutamine synthetase expression by astrocytes suggesting a neuroprotective role in the ageing brain. *Neuroscience Letters*, **2015**, 609, 11-7	3.3	34
244	The widening spectrum of C9ORF72-related disease; genotype/phenotype correlations and potential modifiers of clinical phenotype. *Acta Neuropathologica*, **2014**, 127, 333-45	14.3	124
243	TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. *Acta Neuropathologica*, **2014**, 127, 407-18	14.3	97
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