# Pamela J Shaw

List of Publications by Year
in descending order

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|---|---|---|---|
| 322 papers | 26,361 citations | 6254 80 h-index | 8866 145 g-index |
| 334 all docs | 334 docs citations | 334 times ranked | 22903 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Assessment of the Precision in Measuring Glutathione at <scp>3â€‰T</scp> With a <scp>MEGAâ€PRESS</scp> Sequence in Primary Motor Cortex and Occipital Cortex. Journal of Magnetic Resonance Imaging, 2022, 55, 435-442. | 3.4 | 2 |
| 2 | Reinnervation as measured by the motor unit size index is associated with preservation of muscle strength in amyotrophic lateral sclerosis, but not all muscles reinnervate. Muscle and Nerve, 2022, 65, 203-210. | 2.2 | 6 |
| 3 | The application of Raman spectroscopy to the diagnosis of mitochondrial muscle disease: A preliminary comparison between fibre optic probe and microscope formats. Journal of Raman Spectroscopy, 2022, 53, 172-181. | 2.5 | 5 |
| 4 | A review of Mendelian randomization in amyotrophic lateral sclerosis. Brain, 2022, 145, 832-842. | 7.6 | 29 |
| 5 | Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. Neuron, 2022, 110, 992-1008.e11. | 8.1 | 51 |
| 6 | Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8. | 3.8 | 23 |
| 7 | Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029. | 3.3 | 29 |
| 8 | Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264. | 12.4 | 38 |
| 9 | Unbiased metabolome screen leads to personalized medicine strategy for amyotrophic lateral sclerosis. Brain Communications, 2022, 4, fcac069. | 3.3 | 10 |
| 10 | SPG15 protein deficits are at the crossroads between lysosomal abnormalities, altered lipid metabolism and synaptic dysfunction. Human Molecular Genetics, 2022, 31, 2693-2710. | 2.9 | 6 |
| 11 | Whole-genome sequencing reveals that variants in the Interleukin 18 Receptor Accessory Protein 3â€²UTR protect against ALS. Nature Neuroscience, 2022, 25, 433-445. | 14.8 | 16 |
| 12 | Neurotoxic Astrocytes Directly Converted from Sporadic and Familial ALS Patient Fibroblasts Reveal Signature Diversities and miR-146a Theragnostic Potential in Specific Subtypes. Cells, 2022, 11, 1186. | 4.1 | 11 |
| 13 | Rapid identification of human muscle disease with fibre optic Raman spectroscopy. Analyst, The, 2022, 147, 2533-2540. | 3.5 | 9 |
| 14 | Tensor electrical impedance myography identifies bulbar disease progression in amyotrophic lateral sclerosis. Clinical Neurophysiology, 2022, 139, 69-75. | 1.5 | 3 |
| 15 | Creatine kinase and prognosis in amyotrophic lateral sclerosis: a literature review and multi-centre cohort analysis. Journal of Neurology, 2022, 269, 5395-5404. | 3.6 | 6 |
| 16 | Simultaneous ALS and SCA2 associated with an intermediate-length ATXN2 CAG-repeat expansion. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 579-582. | 1.7 | 13 |
| 17 | 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. | 6.7 | 31 |
| 18 | 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. | 1.7 | 2 |

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|---|---|---|---|
| 19 | The Effect of <scp>SMN</scp> Gene Dosage on <scp>ALS</scp> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697. | 5.3 | 10 |
| 20 | The gut microbiome: a key player in the complexity of amyotrophic lateral sclerosis (ALS). BMC Medicine, 2021, 19, 13. | 5.5 | 52 |
| 21 | Mitochondrial Dysfunction in Alzheimerâ€™s Disease: A Biomarker of the Future?. Biomedicines, 2021, 9, 63. | 3.2 | 62 |
| 22 | Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518. | 1.9 | 69 |
| 23 | Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90. | 8.8 | 49 |
| 24 | In Vivo Fiber Optic Raman Spectroscopy of Muscle in Preclinical Models of Amyotrophic Lateral Sclerosis and Duchenne Muscular Dystrophy. ACS Chemical Neuroscience, 2021, 12, 1768-1776. | 3.5 | 12 |
| 25 | Adipose-derived stem cells protect motor neurons and reduce glial activation in both inÂvitro and inÂvivo models of ALS. Molecular Therapy - Methods and Clinical Development, 2021, 21, 413-433. | 4.1 | 11 |
| 26 | Proteomic Approaches to Study Cysteine Oxidation: Applications in Neurodegenerative Diseases. Frontiers in Molecular Neuroscience, 2021, 14, 678837. | 2.9 | 10 |
| 27 | Physical exercise is a risk factor for amyotrophic lateral sclerosis: Convergent evidence from Mendelian randomisation, transcriptomics and risk genotypes. EBioMedicine, 2021, 68, 103397. | 6.1 | 65 |
| 28 | Innovating Clinical Trials for Amyotrophic Lateral Sclerosis. Neurology, 2021, 97, 528-536. | 1.1 | 19 |
| 29 | SRSF1-dependent inhibition of C9ORF72-repeat RNA nuclear export: genome-wide mechanisms for neuroprotection in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2021, 16, 53. | 10.8 | 13 |
| 30 | Extensive phenotypic characterisation of a human TDP-43Q331KÂtransgenic mouse model of amyotrophic lateral sclerosis (ALS). Scientific Reports, 2021, 11, 16659. | 3.3 | 12 |
| 31 | Tensor electrical impedance myography identifies clinically relevant features in amyotrophic lateral sclerosis. Physiological Measurement, 2021, 42, 105004. | 2.1 | 4 |
| 32 | Amyotrophic lateral sclerosis alters the metabolic aging profile in patient derived fibroblasts. Neurobiology of Aging, 2021, 105, 64-77. | 3.1 | 16 |
| 33 | Amyotrophic lateral sclerosis transcriptomics reveals immunological effects of low-dose interleukin-2. Brain Communications, 2021, 3, fcab141. | 3.3 | 17 |
| 34 | 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. | 5.2 | 17 |
| 35 | Membrane lipid raft homeostasis is directly linked to neurodegeneration. Essays in Biochemistry, 2021, 65, 999-1011. | 4.7 | 15 |
| 36 | Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648. | 21.4 | 223 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Proteinopathies as Hallmarks of Impaired Gene Expression, Proteostasis and Mitochondrial Function in Amyotrophic Lateral Sclerosis. Frontiers in Neuroscience, 2021, 15, 783624. | 2.8 | 13 |
| 38 | Longitudinal multi-modal muscle-based biomarker assessment in motor neuron disease. Journal of Neurology, 2020, 267, 244-256. | 3.6 | 15 |
| 39 | Disrupted glycosylation of lipids and proteins is a cause of neurodegeneration. Brain, 2020, 143, 1332-1340. | 7.6 | 58 |
| 40 | Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. Cell Reports, 2020, 33, 108456. | 6.4 | 24 |
| 41 | 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. | 1.7 | 7 |
| 42 | 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. | 6.1 | 41 |
| 43 | SOD1-targeting therapies for neurodegenerative diseases: a review of current findings and future potential. Expert Opinion on Orphan Drugs, 2020, 8, 379-392. | 0.8 | 2 |
| 44 | Biomarkers in amyotrophic lateral sclerosis: a review of new developments. Current Opinion in Neurology, 2020, 33, 662-668. | 3.6 | 20 |
| 45 | Deficits in Mitochondrial Spare Respiratory Capacity Contribute to the Neuropsychological Changes of Alzheimerâ€™s Disease. Journal of Personalized Medicine, 2020, 10, 32. | 2.5 | 20 |
| 46 | Oligodendrocyte pathology exceeds axonal pathology in white matter in human amyotrophic lateral sclerosis. Journal of Pathology, 2020, 251, 262-271. | 4.5 | 37 |
| 47 | Phase 1â€“2 Trial of Antisense Oligonucleotide Tofersen for SOD1 ALS. New England Journal of Medicine, 2020, 383, 109-119. | 27.0 | 354 |
| 48 | Multi-dimensional electrical impedance myography of the tongue as a potential biomarker for amyotrophic lateral sclerosis. Clinical Neurophysiology, 2020, 131, 799-808. | 1.5 | 16 |
| 49 | UK case control study of smoking and risk of amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 222-227. | 1.7 | 10 |
| 50 | Modelling and analysis of electrical impedance myography of the lateral tongue. Physiological Measurement, 2020, 41, 125008. | 2.1 | 6 |
| 51 | Magnetic resonance spectroscopy reveals mitochondrial dysfunction in amyotrophic lateral sclerosis. Brain, 2020, 143, 3603-3618. | 7.6 | 24 |
| 52 | ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. Brain Communications, 2020, 2, fcaa064. | 3.3 | 33 |
| 53 | Cross-reactive probes on Illumina DNA methylation arrays: a large study on ALS shows that a cautionary approach is warranted in interpreting epigenome-wide association studies. NAR Genomics and Bioinformatics, 2020, 2, lqaa105. | 3.2 | 13 |
| 54 | C9orf72 intermediate expansions of 24â€“30 repeats are associated with ALS. Acta Neuropathologica Communications, 2019, 7, 115. | 5.2 | 75 |

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|---|---|---|---|
| 55 | C9orf72 expansion within astrocytes reduces metabolic flexibility in amyotrophic lateral sclerosis. Brain, 2019, 142, 3771-3790. | 7.6 | 59 |
| 56 | Astrocyte adenosine deaminase loss increases motor neuron toxicity in amyotrophic lateral sclerosis. Brain, 2019, 142, 586-605. | 7.6 | 84 |
| 57 | Telomere length is greater in ALS than in controls: a whole genome sequencing study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 229-234. | 1.7 | 18 |
| 58 | Biomarkers in Motor Neuron Disease: A State of the Art Review. Frontiers in Neurology, 2019, 10, 291. | 2.4 | 87 |
| 59 | Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481. | 5.3 | 118 |
| 60 | Mutations in the Glycosyltransferase Domain of GLT8D1 Are Associated with Familial Amyotrophic Lateral Sclerosis. Cell Reports, 2019, 26, 2298-2306.e5. | 6.4 | 57 |
| 61 | Using telehealth in motor neuron disease to increase access to specialist multidisciplinary care: a UK-based pilot and feasibility study. BMJ Open, 2019, 9, e028525. | 1.9 | 20 |
| 62 | Process evaluation and exploration of telehealth in motor neuron disease in a UK specialist centre. BMJ Open, 2019, 9, e028526. | 1.9 | 22 |
| 63 | Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. Science Translational Medicine, 2019, 11, . | 12.4 | 37 |
| 64 | Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974. | 14.8 | 101 |
| 65 | Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. Neurobiology of Aging, 2019, 74, 234.e9-234.e15. | 3.1 | 26 |
| 66 | Striking phenotypic variation in a family with the P506S UBQLN2 mutation including amyotrophic lateral sclerosis, spastic paraplegia, and frontotemporal dementia. Neurobiology of Aging, 2019, 73, 229.e5-229.e9. | 3.1 | 16 |
| 67 | Younger age of onset in familial amyotrophic lateral sclerosis is a result of pathogenic gene variants, rather than ascertainment bias. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 268-271. | 1.9 | 38 |
| 68 | The role of mitochondria in amyotrophic lateral sclerosis. Neuroscience Letters, 2019, 710, 132933. | 2.1 | 356 |
| 69 | 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 |
| 70 | Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6. | 8.1 | 517 |
| 71 | 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. | 1.7 | 37 |
| 72 | Imaging muscle as a potential biomarker of denervation in motor neuron disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 248-255. | 1.9 | 41 |

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|---|---|---|---|
| 73 | Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3. | 1.7 | 22 |
| 74 | Lost in translation: microRNAs mediate pathological cross-talk between motor neurons and astrocytes. Brain, 2018, 141, 2534-2536. | 7.6 | 4 |
| 75 | Stable transgenic C9orf72 zebrafish model key aspects of the ALS/FTD phenotype and reveal novel pathological features. Acta Neuropathologica Communications, 2018, 6, 125. | 5.2 | 47 |
| 76 | Novel genotype-phenotype and MRI correlations in a large cohort of patients with SPG7 mutations. Neurology: Genetics, 2018, 4, e279. | 1.9 | 44 |
| 77 | 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. | 4.2 | 63 |
| 78 | Efficacy of the Head Up collar in facilitating functional head movements in patients with Amyotrophic Lateral Sclerosis. Clinical Biomechanics, 2018, 57, 114-120. | 1.2 | 7 |
| 79 | 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. | 5.1 | 33 |
| 80 | C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 281.1-281. | 1.9 | 33 |
| 81 | â€œAnything that makes lifeâ€™s journey better.â€•Exploring the use of digital technology by people living with motor neurone disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 378-387. | 1.7 | 15 |
| 82 | Comparison of the Kingâ€™s and MiToS staging systems for ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 227-232. | 1.7 | 58 |
| 83 | Serum miRNAs miR-206, 143-3p and 374b-5p as potential biomarkers for amyotrophic lateral sclerosis (ALS). Neurobiology of Aging, 2017, 55, 123-131. | 3.1 | 117 |
| 84 | A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. Brain, 2017, 140, 1611-1618. | 7.6 | 71 |
| 85 | Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, . | 12.4 | 129 |
| 86 | Viral delivery of C9ORF72 hexanucleotide repeat expansions in mice lead to repeat length dependent neuropathology and behavioral deficits.. DMM Disease Models and Mechanisms, 2017, 10, 859-868. | 2.4 | 25 |
| 87 | A data-driven approach links microglia to pathology and prognosis in amyotrophic lateral sclerosis. Acta Neuropathologica Communications, 2017, 5, 23. | 5.2 | 63 |
| 88 | Amyotrophic lateral sclerosis. Nature Reviews Disease Primers, 2017, 3, 17071. | 30.5 | 885 |
| 89 | Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922. | 1.1 | 82 |
| 90 | Can Astrocytes Be a Target for Precision Medicine?. Advances in Experimental Medicine and Biology, 2017, 1007, 111-128. | 1.6 | 7 |

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|---|---|---|---|
| 91 | Gene Therapy in the Nervous System: Failures and Successes. Advances in Experimental Medicine and Biology, 2017, 1007, 241-257. | 1.6 | 6 |
| 92 | Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903. | 5.5 | 277 |
| 93 | C9orf72 expansion disrupts ATM-mediated chromosomal break repair. Nature Neuroscience, 2017, 20, 1225-1235. | 14.8 | 138 |
| 94 | SRSF1-dependent nuclear export inhibition of C9ORF72 repeat transcripts prevents neurodegeneration and associated motor deficits. Nature Communications, 2017, 8, 16063. | 12.8 | 106 |
| 95 | 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 |
| 96 | Protein Homeostasis in Amyotrophic Lateral Sclerosis: Therapeutic Opportunities?. Frontiers in Molecular Neuroscience, 2017, 10, 123. | 2.9 | 62 |
| 97 | Targeted Genetic Screen in Amyotrophic Lateral Sclerosis Reveals Novel Genetic Variants with Synergistic Effect on Clinical Phenotype. Frontiers in Molecular Neuroscience, 2017, 10, 370. | 2.9 | 24 |
| 98 | C9ORF72 hexanucleotide repeat exerts toxicity in a stable, inducible motor neuronal cell model, which is rescued by partial depletion of Pten. Human Molecular Genetics, 2017, 26, 1133-1145. | 2.9 | 23 |
| 99 | Advances, challenges and future directions for stem cell therapy in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2017, 12, 85. | 10.8 | 51 |
| 100 | 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. | 2.8 | 83 |
| 101 | An Objective Functional Characterisation of Head Movement Impairment in Individuals with Neck Muscle Weakness Due to Amyotrophic Lateral Sclerosis. PLoS ONE, 2017, 12, e0169019. | 2.5 | 8 |
| 102 | Immune response in peripheral axons delays disease progression in SOD1G93A mice. Journal of Neuroinflammation, 2016, 13, 261. | 7.2 | 63 |
| 103 | MicroNeurotrophins Improve Survival in Motor Neuron-Astrocyte Co-Cultures but Do Not Improve Disease Phenotypes in a Mutant SOD1 Mouse Model of Amyotrophic Lateral Sclerosis. PLoS ONE, 2016, 11, e0164103. | 2.5 | 18 |
| 104 | Oligogenic inheritance of optineurin (OPTN) and C9ORF72 mutations in ALS highlights localisation of OPTN in the TDPâ€43â€negative inclusions of C9ORF72â€ALS. Neuropathology, 2016, 36, 125-134. | 1.2 | 35 |
| 105 | Motor neurone disease/amyotrophic lateral sclerosis associated with intermediateâ€length <scp>CAG</scp> repeat expansions in <scp>Ataxinâ€2</scp> does not have 1<scp>C</scp>2â€positive polyglutamine inclusions. Neuropathology and Applied Neurobiology, 2016, 42, 377-389. | 3.2 | 7 |
| 106 | The role of cranial and thoracic electromyography within diagnostic criteria for amyotrophic lateral sclerosis. Muscle and Nerve, 2016, 54, 378-385. | 2.2 | 6 |
| 107 | The C9orf72 protein interacts with Rab1a and the <scp>ULK</scp>1 complex to regulate initiation of autophagy. EMBO Journal, 2016, 35, 1656-1676. | 7.8 | 327 |
| 108 | Using technology to improve access to specialist care in amyotrophic lateral sclerosis: A systematic review. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 313-324. | 1.7 | 35 |

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|---|---|---|---|
| 109 | Assessment of the Sheffield Support Snood, an innovative cervical orthosis designed for people affected by neck muscle weakness. Clinical Biomechanics, 2016, 32, 201-206. | 1.2 | 21 |
| 110 | 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, 436-442. | 1.7 | 27 |
| 111 | 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. | 1.7 | 2 |
| 112 | Oligodendrocytes contribute to motor neuron death in ALS via SOD1-dependent mechanism. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E6496-E6505. | 7.1 | 139 |
| 113 | Clinical aspects of motor neurone disease. Medicine, 2016, 44, 552-556. | 0.4 | 13 |
| 114 | Neuronal <scp>DNA</scp> damage responseâ€associated dysregulation of signalling pathways and cholesterol metabolism at the earliest stages of <scp>A</scp>lzheimerâ€type pathology. Neuropathology and Applied Neurobiology, 2016, 42, 167-179. | 3.2 | 28 |
| 115 | Rare genetic variation in UNC13A may modify survival in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 593-599. | 1.7 | 22 |
| 116 | Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048. | 21.4 | 494 |
| 117 | NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042. | 21.4 | 218 |
| 118 | Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812. | 9.0 | 57 |
| 119 | Long-term physical activity: an exogenous risk factor for sporadic amyotrophic lateral sclerosis?. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 377-384. | 1.7 | 46 |
| 120 | The impact of gastrostomy in motor neurone disease: challenges and benefits from a patient and carer perspective. BMJ Supportive and Palliative Care, 2016, 6, 52-59. | 1.6 | 33 |
| 121 | DiPALS: Diaphragm Pacing in patients with Amyotrophic Lateral Sclerosis â€“ a randomised controlled trial. Health Technology Assessment, 2016, 20, 1-186. | 2.8 | 13 |
| 122 | Head-Up; An interdisciplinary, participatory and co-design process informing the development of a novel head and neck support for people living with progressive neck muscle weakness. Journal of Medical Engineering and Technology, 2015, 39, 404-410. | 1.4 | 25 |
| 123 | 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. | 3.7 | 36 |
| 124 | A Reduced Astrocyte Response to Î²-Amyloid Plaques in the Ageing Brain Associates with Cognitive Impairment. PLoS ONE, 2015, 10, e0118463. | 2.5 | 45 |
| 125 | C9ORF72 GGGGCC Expanded Repeats Produce Splicing Dysregulation which Correlates with Disease Severity in Amyotrophic Lateral Sclerosis. PLoS ONE, 2015, 10, e0127376. | 2.5 | 83 |
| 126 | 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, 11-17. | 2.1 | 58 |

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|---|---|---|---|
| 127 | The Spectrum of C9orf72-mediated Neurodegeneration and Amyotrophic Lateral Sclerosis. Neurotherapeutics, 2015, 12, 326-339. | 4.4 | 46 |
| 128 | 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, 108-113. | 1.7 | 7 |
| 129 | Intermediate length C9orf72 expansion in an ALS patient without classical C9orf72 neuropathology. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 249-251. | 1.7 | 8 |
| 130 | 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, 478-484. | 1.7 | 45 |
| 131 | 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, 448-455. | 1.7 | 39 |
| 132 | Regionality of disease progression predicts prognosis in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 442-447. | 1.7 | 3 |
| 133 | Stratified gene expression analysis identifies major amyotrophic lateral sclerosis genes. Neurobiology of Aging, 2015, 36, 2006.e1-2006.e9. | 3.1 | 22 |
| 134 | Antisense RNA foci in the motor neurons of C9ORF72-ALS patients are associated with TDP-43 proteinopathy. Acta Neuropathologica, 2015, 130, 63-75. | 7.7 | 149 |
| 135 | Current developments in gene therapy for amyotrophic lateral sclerosis. Expert Opinion on Biological Therapy, 2015, 15, 935-947. | 3.1 | 30 |
| 136 | The role of TREM2 R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. Alzheimer's and Dementia, 2015, 11, 1407-1416. | 0.8 | 152 |
| 137 | Altered age-related changes in bioenergetic properties and mitochondrial morphology in fibroblasts from sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2015, 36, 2893-2903. | 3.1 | 38 |
| 138 | PTEN Depletion Decreases Disease Severity and Modestly Prolongs Survival in a Mouse Model of Spinal Muscular Atrophy. Molecular Therapy, 2015, 23, 270-277. | 8.2 | 47 |
| 139 | A neuronal <scp>DNA</scp> damage response is detected at the earliest stages of <scp>A</scp>lzheimer's neuropathology and correlates with cognitive impairment in the <scp>M</scp>edical <scp>R</scp>esearch <scp>C</scp>ouncil's <scp>C</scp>ognitive <scp>F</scp>unction and <scp>A</scp>geing <scp>S</scp>tudy ageing brain cohort. Neuropathology and Applied Neurobiology, 2015, 41, 483-496. | 3.2 | 40 |
| 140 | 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. | 3.1 | 63 |
| 141 | Use of clinical staging in amyotrophic lateral sclerosis for phase 3 clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 45-49. | 1.9 | 75 |
| 142 | Gene expression signatures in motor neurone disease fibroblasts reveal dysregulation of metabolism, hypoxiaâ€response and <scp>RNA</scp> processing functions. Neuropathology and Applied Neurobiology, 2015, 41, 201-226. | 3.2 | 73 |
| 143 | Invited Review: Decoding the pathophysiological mechanisms that underlie <scp>RNA</scp> dysregulation in neurodegenerative disorders: a review of the current state of the art. Neuropathology and Applied Neurobiology, 2015, 41, 109-134. | 3.2 | 47 |
| 144 | A new zebrafish model produced by TILLING of SOD1-related amyotrophic lateral sclerosis replicates key features of the disease and represents a tool for in vivo therapeutic screening. DMM Disease Models and Mechanisms, 2014, 7, 73-81. | 2.4 | 53 |

| # | Article | IF | Citations |
|---|---|---|---|
| 145 | A zebrafish model exemplifies the long preclinical period of motor neuron disease. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1288-1289. | 1.9 | 8 |
| 146 | Direct conversion of patient fibroblasts demonstrates non-cell autonomous toxicity of astrocytes to motor neurons in familial and sporadic ALS. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 829-832. | 7.1 | 296 |
| 147 | Factors influencing decision-making in relation to timing of gastrostomy insertion in patients with motor neurone disease. BMJ Supportive and Palliative Care, 2014, 4, 57-63. | 1.6 | 43 |
| 148 | Sequestration of multiple RNA recognition motif-containing proteins by C9orf72 repeat expansions. Brain, 2014, 137, 2040-2051. | 7.6 | 253 |
| 149 | The widening spectrum of C9ORF72-related disease; genotype/phenotype correlations and potential modifiers of clinical phenotype. Acta Neuropathologica, 2014, 127, 333-345. | 7.7 | 150 |
| 150 | TMEM106B is a genetic modifier of frontotemporal lobar degeneration with C9orf72 hexanucleotide repeat expansions. Acta Neuropathologica, 2014, 127, 407-418. | 7.7 | 123 |
| 151 | Superoxide dismutase 1 mutation in a cellular model of amyotrophic lateral sclerosis shifts energy generation from oxidative phosphorylation to glycolysis. Neurobiology of Aging, 2014, 35, 1499-1509. | 3.1 | 77 |
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