

Claire Troakes

List of Publications by Year in descending order

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Version: 2024-02-01

104
papers

9,754
citations

47006

47
h-index

42399

92
g-index

109
all docs

109
docs citations

109
times ranked

13890
citing authors

| # | ARTICLE | IF | CITATIONS |
|----|---|------|-----------|
| 1 | Clinical diagnosis of progressive supranuclear palsy: The movement disorder society criteria. <i>Movement Disorders</i> , 2017, 32, 853-864. | 3.9 | 1,402 |
| 2 | Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6. | 8.1 | 517 |
| 3 | Methylomic profiling implicates cortical deregulation of ANK1 in Alzheimer's disease. <i>Nature Neuroscience</i> , 2014, 17, 1164-1170. | 14.8 | 488 |
| 4 | Hexanucleotide Repeats in ALS/FTD Form Length-Dependent RNA Foci, Sequester RNA Binding Proteins, and Are Neurotoxic. <i>Cell Reports</i> , 2013, 5, 1178-1186. | 6.4 | 419 |
| 5 | The genetics and neuropathology of amyotrophic lateral sclerosis. <i>Acta Neuropathologica</i> , 2012, 124, 339-352. | 7.7 | 346 |
| 6 | Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331. | 8.1 | 308 |
| 7 | Methylation QTLs in the developing brain and their enrichment in schizophrenia risk loci. <i>Nature Neuroscience</i> , 2016, 19, 48-54. | 14.8 | 306 |
| 8 | Plasma p-tau231: a new biomarker for incipient Alzheimer's disease pathology. <i>Acta Neuropathologica</i> , 2021, 141, 709-724. | 7.7 | 285 |
| 9 | NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042. | 21.4 | 218 |
| 10 | Distribution patterns of tau pathology in progressive supranuclear palsy. <i>Acta Neuropathologica</i> , 2020, 140, 99-119. | 7.7 | 210 |
| 11 | Plasma p-tau181 accurately predicts Alzheimer's disease pathology at least 8 years prior to post-mortem and improves the clinical characterisation of cognitive decline. <i>Acta Neuropathologica</i> , 2020, 140, 267-278. | 7.7 | 209 |
| 12 | The C9ORF72 expansion mutation is a common cause of ALS+/FTD in Europe and has a single founder. <i>European Journal of Human Genetics</i> , 2013, 21, 102-108. | 2.8 | 201 |
| 13 | Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. <i>Nature Genetics</i> , 2021, 53, 294-303. | 21.4 | 198 |
| 14 | Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. <i>Lancet Neurology</i> , The, 2018, 17, 64-74. | 10.2 | 195 |
| 15 | Aluminium in brain tissue in familial Alzheimer's disease. <i>Journal of Trace Elements in Medicine and Biology</i> , 2017, 40, 30-36. | 3.0 | 182 |
| 16 | Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. <i>Human Molecular Genetics</i> , 2014, 23, 6139-6146. | 2.9 | 178 |
| 17 | Elevated DNA methylation across a 48 kb region spanning the HOXA gene cluster is associated with Alzheimer's disease neuropathology. <i>Alzheimer's and Dementia</i> , 2018, 14, 1580-1588. | 0.8 | 138 |
| 18 | A histone acetylome-wide association study of Alzheimer's disease identifies disease-associated H3K27ac differences in the entorhinal cortex. <i>Nature Neuroscience</i> , 2018, 21, 1618-1627. | 14.8 | 138 |

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|----|--|------|-----------|
| 19 | Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017, 9, . | 12.4 | 129 |
| 20 | Increased plasma neurofilament light chain concentration correlates with severity of post-mortem neurofibrillary tangle pathology and neurodegeneration. <i>Acta Neuropathologica Communications</i> , 2019, 7, 5. | 5.2 | 125 |
| 21 | Which ante mortem clinical features predict progressive supranuclear palsy pathology?. <i>Movement Disorders</i> , 2017, 32, 995-1005. | 3.9 | 121 |
| 22 | In vitro prion-like behaviour of TDP-43 in ALS. <i>Neurobiology of Disease</i> , 2016, 96, 236-247. | 4.4 | 118 |
| 23 | Cross-region reduction in 5-hydroxymethylcytosine in Alzheimer's disease brain. <i>Neurobiology of Aging</i> , 2014, 35, 1850-1854. | 3.1 | 114 |
| 24 | An MND/ALS phenotype associated with <i>C9orf72</i> repeat expansion: Abundant p62 ⁺ positive, TDP ⁺ negative inclusions in cerebral cortex, hippocampus and cerebellum but without associated cognitive decline. <i>Neuropathology</i> , 2012, 32, 505-514. | 1.2 | 110 |
| 25 | Upregulation of calpain activity precedes tau phosphorylation and loss of synaptic proteins in Alzheimer's disease brain. <i>Acta Neuropathologica Communications</i> , 2016, 4, 34. | 5.2 | 100 |
| 26 | Invited Review: The spectrum of neuropathology in COVID-19. <i>Neuropathology and Applied Neurobiology</i> , 2021, 47, 3-16. | 3.2 | 99 |
| 27 | Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. <i>Lancet Neurology</i> , The, 2018, 17, 548-558. | 10.2 | 97 |
| 28 | How to apply the movement disorder society criteria for diagnosis of progressive supranuclear palsy. <i>Movement Disorders</i> , 2019, 34, 1228-1232. | 3.9 | 93 |
| 29 | Genome-wide analyses as part of the international FTL-D-TDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTL. <i>Acta Neuropathologica</i> , 2019, 137, 879-899. | 7.7 | 90 |
| 30 | Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. <i>Neurobiology of Aging</i> , 2015, 36, 1602.e17-1602.e27. | 3.1 | 87 |
| 31 | ATXN2 trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9. | 3.1 | 86 |
| 32 | Parallel profiling of DNA methylation and hydroxymethylation highlights neuropathology-associated epigenetic variation in Alzheimer's disease. <i>Clinical Epigenetics</i> , 2019, 11, 52. | 4.1 | 84 |
| 33 | Variation in 5-hydroxymethylcytosine across human cortex and cerebellum. <i>Genome Biology</i> , 2016, 17, 27. | 8.8 | 83 |
| 34 | 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 |
| 35 | Dipeptide repeat protein inclusions are rare in the spinal cord and almost absent from motor neurons in C9ORF72 mutant amyotrophic lateral sclerosis and are unlikely to cause their degeneration. <i>Acta Neuropathologica Communications</i> , 2015, 3, 38. | 5.2 | 80 |
| 36 | Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. <i>Neurobiology of Aging</i> , 2016, 38, 214.e7-214.e10. | 3.1 | 78 |

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|----|--|-----|-----------|
| 37 | Amyotrophic lateral sclerosis-like superoxide dismutase 1 proteinopathy is associated with neuronal loss in Parkinson's disease brain. <i>Acta Neuropathologica</i> , 2017, 134, 113-127. | 7.7 | 78 |
| 38 | A feedback loop between dipeptide-repeat protein, TDP-43 and karyopherin- β mediates C9orf72-related neurodegeneration. <i>Brain</i> , 2018, 141, 2908-2924. | 7.6 | 75 |
| 39 | Schizophrenia-associated methylomic variation: molecular signatures of disease and polygenic risk burden across multiple brain regions. <i>Human Molecular Genetics</i> , 2017, 26, ddw373. | 2.9 | 74 |
| 40 | Transcriptomic analysis of probable asymptomatic and symptomatic alzheimer brains. <i>Brain, Behavior, and Immunity</i> , 2019, 80, 644-656. | 4.1 | 72 |
| 41 | Genome-wide DNA methylation profiling identifies convergent molecular signatures associated with idiopathic and syndromic autism in post-mortem human brain tissue. <i>Human Molecular Genetics</i> , 2019, 28, 2201-2211. | 2.9 | 70 |
| 42 | C9orf72 poly GA RAN-translated protein plays a key role in amyotrophic lateral sclerosis via aggregation and toxicity. <i>Human Molecular Genetics</i> , 2017, 26, 4765-4777. | 2.9 | 64 |
| 43 | Neurodegeneration in frontotemporal lobar degeneration and motor neurone disease associated with expansions in <i>C9orf72</i> is linked to TDP-43 pathology and not associated with aggregated forms of dipeptide repeat proteins. <i>Neuropathology and Applied Neurobiology</i> , 2016, 42, 242-254. | 3.2 | 61 |
| 44 | A cross-brain regions study of ANK1 DNA methylation in different neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2019, 74, 70-76. | 3.1 | 58 |
| 45 | On the identification of low allele frequency mosaic mutations in the brains of Alzheimer's disease patients. <i>Alzheimer's and Dementia</i> , 2015, 11, 1265-1276. | 0.8 | 57 |
| 46 | Neuropathology of the hippocampus in FTD-Tau with Pick bodies: a study of the BrainNet Europe Consortium. <i>Neuropathology and Applied Neurobiology</i> , 2013, 39, 166-178. | 3.2 | 54 |
| 47 | Epigenomic and transcriptomic signatures of a Klinefelter syndrome (47,XXY) karyotype in the brain. <i>Epigenetics</i> , 2014, 9, 587-599. | 2.7 | 53 |
| 48 | Proteomic analyses reveal that loss of TDP-43 affects RNA processing and intracellular transport. <i>Neuroscience</i> , 2015, 293, 157-170. | 2.3 | 52 |
| 49 | Alzheimer-related decrease in CYFIP2 links amyloid production to tau hyperphosphorylation and memory loss. <i>Brain</i> , 2016, 139, 2751-2765. | 7.6 | 52 |
| 50 | C9orf72 intermediate repeats are associated with corticobasal degeneration, increased C9orf72 expression and disruption of autophagy. <i>Acta Neuropathologica</i> , 2019, 138, 795-811. | 7.7 | 50 |
| 51 | The Identification of Aluminum in Human Brain Tissue Using Lumogallion and Fluorescence Microscopy. <i>Journal of Alzheimer's Disease</i> , 2016, 54, 1333-1338. | 2.6 | 48 |
| 52 | Copathology in Progressive Supranuclear Palsy: Does It Matter?. <i>Movement Disorders</i> , 2020, 35, 984-993. | 3.9 | 48 |
| 53 | Retention of hexanucleotide repeat-containing intron in C9orf72 mRNA: implications for the pathogenesis of ALS/FTD. <i>Acta Neuropathologica Communications</i> , 2016, 4, 18. | 5.2 | 46 |
| 54 | 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 |

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|----|---|------|-----------|
| 55 | The Increased Densities, But Different Distributions, of Both C3 and S100A10 Immunopositive Astrocyte-Like Cells in Alzheimer's Disease Brains Suggest Possible Roles for Both A1 and A2 Astrocytes in the Disease Pathogenesis. <i>Brain Sciences</i> , 2020, 10, 503. | 2.3 | 43 |
| 56 | The Psychiatric Risk Gene NT5C2 Regulates Adenosine Monophosphate-Activated Protein Kinase Signaling and Protein Translation in Human Neural Progenitor Cells. <i>Biological Psychiatry</i> , 2019, 86, 120-130. | 1.3 | 42 |
| 57 | Genome-wide significant schizophrenia risk variation on chromosome 10q24 is associated with altered cis-regulation of BORCS7, AS3MT, and NT5C2 in the human brain. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2016, 171, 806-814. | 1.7 | 41 |
| 58 | Disruption of endoplasmic reticulum-mitochondria tethering proteins in post-mortem Alzheimer's disease brain. <i>Neurobiology of Disease</i> , 2020, 143, 105020. | 4.4 | 41 |
| 59 | ABCA7 p.G215S as potential protective factor for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 46, 235.e1-235.e9. | 3.1 | 37 |
| 60 | Validation of the Movement Disorder Society Criteria for the Diagnosis of 4-Repeat Tauopathies. <i>Movement Disorders</i> , 2020, 35, 171-176. | 3.9 | 37 |
| 61 | Symmetric dimethylation of poly-GR correlates with disease duration in C9orf72 FTL and ALS and reduces poly-GR phase separation and toxicity. <i>Acta Neuropathologica</i> , 2020, 139, 407-410. | 7.7 | 36 |
| 62 | Evidence that the presynaptic vesicle protein CSPalpha is a key player in synaptic degeneration and protection in Alzheimer's disease. <i>Molecular Brain</i> , 2015, 8, 6. | 2.6 | 34 |
| 63 | Tau deposition patterns are associated with functional connectivity in primary tauopathies. <i>Nature Communications</i> , 2022, 13, 1362. | 12.8 | 34 |
| 64 | Comparison of clinical and neuropathological diagnoses of neurodegenerative diseases in two centres from the Brains for Dementia Research (BDR) cohort. <i>Journal of Neural Transmission</i> , 2019, 126, 327-337. | 2.8 | 33 |
| 65 | Tissue-specific patterns of allelically-skewed DNA methylation. <i>Epigenetics</i> , 2016, 11, 24-35. | 2.7 | 32 |
| 66 | Heritability and genetic variance of dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2019, 127, 492-501. | 4.4 | 29 |
| 67 | Exome sequencing identifies 2 novel presenilin 1 mutations (p.L166V and p.S230R) in British early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014, 35, 2422.e13-2422.e16. | 3.1 | 28 |
| 68 | Transportin 1 colocalization with Fused in Sarcoma (FUS) inclusions is not characteristic for amyotrophic lateral sclerosis-FUS confirming disrupted nuclear import of mutant FUS and distinguishing it from frontotemporal lobar degeneration with FUS inclusions. <i>Neuropathology and Applied Neurobiology</i> , 2013, 39, 553-561. | 3.2 | 27 |
| 69 | Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. <i>Acta Neuropathologica Communications</i> , 2020, 8, 5. | 5.2 | 27 |
| 70 | Clusterin expression is upregulated following acute head injury and localizes to astrocytes in old head injury. <i>Neuropathology</i> , 2017, 37, 12-24. | 1.2 | 24 |
| 71 | ALS-FUS pathology revisited: singleton FUS mutations and an unusual case with both a FUS and TARDBP mutation. <i>Acta Neuropathologica Communications</i> , 2015, 3, 62. | 5.2 | 22 |
| 72 | The Neuropathological Diagnosis of Alzheimer's Disease—The Challenges of Pathological Mimics and Concomitant Pathology. <i>Brain Sciences</i> , 2020, 10, 479. | 2.3 | 22 |

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|----|--|-----|-----------|
| 73 | Clinical Conditions “Suggestive of Progressive Supranuclear Palsy” Diagnostic Performance. <i>Movement Disorders</i> , 2020, 35, 2301-2313. | 3.9 | 22 |
| 74 | Gamma-synuclein pathology in amyotrophic lateral sclerosis. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 29-37. | 3.7 | 21 |
| 75 | Frequency and signature of somatic variants in 1461 human brain exomes. <i>Genetics in Medicine</i> , 2019, 21, 904-912. | 2.4 | 20 |
| 76 | The CHCHD10 P34S variant is not associated with ALS in a UK cohort of familial and sporadic patients. <i>Neurobiology of Aging</i> , 2015, 36, 2908.e17-2908.e18. | 3.1 | 19 |
| 77 | Extended post-mortem delay times should not be viewed as a deterrent to the scientific investigation of human brain tissue: a study from the Brains for Dementia Research Network Neuropathology Study Group, UK. <i>Acta Neuropathologica</i> , 2016, 132, 753-755. | 7.7 | 18 |
| 78 | Bridging integrator 1 protein loss in Alzheimer’s disease promotes synaptic tau accumulation and disrupts tau release. <i>Brain Communications</i> , 2020, 2, . | 3.3 | 18 |
| 79 | A comparison of mitochondrial DNA isolation methods in frozen post-mortem human brain tissue” applications for studies of mitochondrial genetics in brain disorders. <i>BioTechniques</i> , 2015, 59, 241-246. | 1.8 | 17 |
| 80 | 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 |
| 81 | Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. <i>Brain</i> , 2022, 145, 1757-1762. | 7.6 | 17 |
| 82 | Assessment of the degree of asymmetry of pathological features in neurodegenerative diseases. What is the significance for brain banks?. <i>Journal of Neural Transmission</i> , 2015, 122, 1499-1508. | 2.8 | 16 |
| 83 | Striking phenotypic variation in a family with the P506S UBQLN2 mutation including amyotrophic lateral sclerosis, spastic paraplegia, and frontotemporal dementia. <i>Neurobiology of Aging</i> , 2019, 73, 229.e5-229.e9. | 3.1 | 16 |
| 84 | Multisystem screening reveals SARS-CoV-2 in neurons of the myenteric plexus and in megakaryocytes. <i>Journal of Pathology</i> , 2022, 257, 198-217. | 4.5 | 16 |
| 85 | Astrocytic CXCL12 motif chemokine ligand-1 mediates β -amyloid-induced synaptotoxicity. <i>Journal of Neuroinflammation</i> , 2021, 18, 306. | 7.2 | 16 |
| 86 | Cytoplasmic TDP-43 is involved in cell fate during stress recovery. <i>Human Molecular Genetics</i> , 2021, 31, 166-175. | 2.9 | 15 |
| 87 | SCFD1 expression quantitative trait loci in amyotrophic lateral sclerosis are differentially expressed. <i>Brain Communications</i> , 2021, 3, fcab236. | 3.3 | 14 |
| 88 | Heterogeneous Nuclear Ribonucleoprotein E2 (hnRNP E2) Is a Component of TDP-43 Aggregates Specifically in the A and C Pathological Subtypes of Frontotemporal Lobar Degeneration. <i>Frontiers in Neuroscience</i> , 2019, 13, 551. | 2.8 | 13 |
| 89 | A comprehensive screening of copy number variability in dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2019, 75, 223.e1-223.e10. | 3.1 | 13 |
| 90 | A HML6 endogenous retrovirus on chromosome 3 is upregulated in amyotrophic lateral sclerosis motor cortex. <i>Scientific Reports</i> , 2021, 11, 14283. | 3.3 | 13 |

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|-----|--|-----|-----------|
| 91 | Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2017, 49, 214.e13-214.e15. | 3.1 | 12 |
| 92 | <i>APOE</i> ϵ 4 is also required in <i>TREM2</i> R47H variant carriers for Alzheimer's disease to develop. <i>Neuropathology and Applied Neurobiology</i> , 2019, 45, 183-186. | 3.2 | 12 |
| 93 | The histone modification H3K4me3 is altered at the <i>ANK1</i> locus in Alzheimer's disease brain. <i>Future Science OA</i> , 2021, 7, FSO665. | 1.9 | 10 |
| 94 | Lack of association between TDP-43 pathology and tau mis-splicing in Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 37, 45-46. | 3.1 | 8 |
| 95 | Unusual neuropathological features and increased brain aluminium in a resident of Camelford, UK. <i>Neuropathology and Applied Neurobiology</i> , 2017, 43, 537-541. | 3.2 | 8 |
| 96 | Spinal cord injury as an indicator of abuse in forensic assessment of abusive head trauma (AHT). <i>International Journal of Legal Medicine</i> , 2021, 135, 1481-1498. | 2.2 | 8 |
| 97 | Frequency and methylation status of selected retrotransposition competent L1 loci in amyotrophic lateral sclerosis. <i>Molecular Brain</i> , 2020, 13, 154. | 2.6 | 7 |
| 98 | No Effect of Genome-Wide Significant Schizophrenia Risk Variation at the <i>DRD2</i> Locus on the Allelic Expression of <i>DRD2</i> in Postmortem Striatum. <i>Molecular Neuropsychiatry</i> , 2019, 5, 212-217. | 2.9 | 4 |
| 99 | Axonal injury is detected by β APP immunohistochemistry in rapid death from head injury following road traffic collision. <i>International Journal of Legal Medicine</i> , 2022, 136, 1321-1339. | 2.2 | 3 |
| 100 | A pathologically confirmed case of combined amyotrophic lateral sclerosis with <i>C9orf72</i> mutation and multiple system atrophy. <i>Neuropathology</i> , 0, , . | 1.2 | 3 |
| 101 | Plasma τ 181 accurately predicts Alzheimer's disease pathology at least 8 years prior to postmortem and improves the clinical characterisation of cognitive decline. <i>Alzheimer's and Dementia</i> , 2020, 16, e047539. | 0.8 | 2 |
| 102 | O3-04-03: CROSS-TISSUE METHYLOMIC PROFILING IN ALZHEIMER'S DISEASE. , 2014, 10, P215-P215. | | 0 |
| 103 | P1-211: Genetic influences on amyloid angiopathy and white matter pathology in familial Alzheimer's disease: A comparison of app and PSEN1 mutations. , 2015, 11, P431-P431. | | 0 |
| 104 | P1-155: PostMortem Brain Tissue Characterisation of Inflammatory and Pathological Hallmarks of Alzheimer's Disease During Disease Progression. <i>Alzheimer's and Dementia</i> , 2016, 12, P462. | 0.8 | 0 |