

# Claire Troakes

## List of Publications by Year in descending order

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Version: 2024-02-01

104  
papers

9,754  
citations

46918

47  
h-index

42291

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.	2.2	1,402
2	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	3.8	517
3	Methylomic profiling implicates cortical deregulation of ANK1 in Alzheimer's disease. <i>Nature Neuroscience</i> , 2014, 17, 1164-1170.	7.1	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.	2.9	419
5	The genetics and neuropathology of amyotrophic lateral sclerosis. <i>Acta Neuropathologica</i> , 2012, 124, 339-352.	3.9	346
6	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	3.8	308
7	Methylation QTLs in the developing brain and their enrichment in schizophrenia risk loci. <i>Nature Neuroscience</i> , 2016, 19, 48-54.	7.1	306
8	Plasma p-tau231: a new biomarker for incipient Alzheimer's disease pathology. <i>Acta Neuropathologica</i> , 2021, 141, 709-724.	3.9	285
9	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	9.4	218
10	Distribution patterns of tau pathology in progressive supranuclear palsy. <i>Acta Neuropathologica</i> , 2020, 140, 99-119.	3.9	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.	3.9	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.	1.4	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.	9.4	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.	4.9	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.	1.5	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.	1.4	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.	0.4	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.	7.1	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, .	5.8	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.	2.4	125
21	Which ante mortem clinical features predict progressive supranuclear palsy pathology?. <i>Movement Disorders</i> , 2017, 32, 995-1005.	2.2	121
22	In vitro prion-like behaviour of TDP-43 in ALS. <i>Neurobiology of Disease</i> , 2016, 96, 236-247.	2.1	118
23	Cross-region reduction in 5-hydroxymethylcytosine in Alzheimer's disease brain. <i>Neurobiology of Aging</i> , 2014, 35, 1850-1854.	1.5	114
24	An MND/ALS phenotype associated with <i>C9orf72</i> repeat expansion: Abundant p62 <sup>+</sup> positive, TDP <sup>+</sup> negative inclusions in cerebral cortex, hippocampus and cerebellum but without associated cognitive decline. <i>Neuropathology</i> , 2012, 32, 505-514.	0.7	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.	2.4	100
26	Invited Review: The spectrum of neuropathology in COVID-19. <i>Neuropathology and Applied Neurobiology</i> , 2021, 47, 3-16.	1.8	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.	4.9	97
28	How to apply the movement disorder society criteria for diagnosis of progressive supranuclear palsy. <i>Movement Disorders</i> , 2019, 34, 1228-1232.	2.2	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.	3.9	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.	1.5	87
31	ATXN2 trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9.	1.5	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.	1.8	84
33	Variation in 5-hydroxymethylcytosine across human cortex and cerebellum. <i>Genome Biology</i> , 2016, 17, 27.	3.8	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.	2.4	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.	2.4	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.	1.5	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.	3.9	78
38	A feedback loop between dipeptide-repeat protein, TDP-43 and karyopherin- $\beta$ mediates C9orf72-related neurodegeneration. <i>Brain</i> , 2018, 141, 2908-2924.	3.7	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.	1.4	74
40	Transcriptomic analysis of probable asymptomatic and symptomatic alzheimer brains. <i>Brain, Behavior, and Immunity</i> , 2019, 80, 644-656.	2.0	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.	1.4	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.	1.4	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.	1.8	61
44	A cross-brain regions study of ANK1 DNA methylation in different neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2019, 74, 70-76.	1.5	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.	0.4	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.	1.8	54
47	Epigenomic and transcriptomic signatures of a Klinefelter syndrome (47,XXY) karyotype in the brain. <i>Epigenetics</i> , 2014, 9, 587-599.	1.3	53
48	Proteomic analyses reveal that loss of TDP-43 affects RNA processing and intracellular transport. <i>Neuroscience</i> , 2015, 293, 157-170.	1.1	52
49	Alzheimer-related decrease in CYFIP2 links amyloid production to tau hyperphosphorylation and memory loss. <i>Brain</i> , 2016, 139, 2751-2765.	3.7	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.	3.9	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.	1.2	48
52	Copathology in Progressive Supranuclear Palsy: Does It Matter?. <i>Movement Disorders</i> , 2020, 35, 984-993.	2.2	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.	2.4	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.	2.4	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.	1.1	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.	0.7	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.	1.1	41
58	Disruption of endoplasmic reticulum-mitochondria tethering proteins in post-mortem Alzheimer's disease brain. <i>Neurobiology of Disease</i> , 2020, 143, 105020.	2.1	41
59	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 46, 235.e1-235.e9.	1.5	37
60	Validation of the Movement Disorder Society Criteria for the Diagnosis of 4-Repeat Tauopathies. <i>Movement Disorders</i> , 2020, 35, 171-176.	2.2	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.	3.9	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.	1.3	34
63	Tau deposition patterns are associated with functional connectivity in primary tauopathies. <i>Nature Communications</i> , 2022, 13, 1362.	5.8	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.	1.4	33
65	Tissue-specific patterns of allelically-skewed DNA methylation. <i>Epigenetics</i> , 2016, 11, 24-35.	1.3	32
66	Heritability and genetic variance of dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2019, 127, 492-501.	2.1	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.	1.5	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.	1.8	27
69	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. <i>Acta Neuropathologica Communications</i> , 2020, 8, 5.	2.4	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.	0.7	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.	2.4	22
72	The Neuropathological Diagnosis of Alzheimer's Disease—The Challenges of Pathological Mimics and Concomitant Pathology. <i>Brain Sciences</i> , 2020, 10, 479.	1.1	22

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73	Clinical Conditions “Suggestive of Progressive Supranuclear Palsy” Diagnostic Performance. <i>Movement Disorders</i> , 2020, 35, 2301-2313.	2.2	22
74	Gamma-synuclein pathology in amyotrophic lateral sclerosis. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 29-37.	1.7	21
75	Frequency and signature of somatic variants in 1461 human brain exomes. <i>Genetics in Medicine</i> , 2019, 21, 904-912.	1.1	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.	1.5	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.	3.9	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, .	1.5	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.	0.8	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.	0.9	17
81	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. <i>Brain</i> , 2022, 145, 1757-1762.	3.7	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.	1.4	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.	1.5	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.	2.1	16
85	Astrocytic CXCL12 motif chemokine ligand-1 mediates $\beta$ -amyloid-induced synaptotoxicity. <i>Journal of Neuroinflammation</i> , 2021, 18, 306.	3.1	16
86	Cytoplasmic TDP-43 is involved in cell fate during stress recovery. <i>Human Molecular Genetics</i> , 2021, 31, 166-175.	1.4	15
87	SCFD1 expression quantitative trait loci in amyotrophic lateral sclerosis are differentially expressed. <i>Brain Communications</i> , 2021, 3, fcab236.	1.5	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.	1.4	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.	1.5	13
90	A HML6 endogenous retrovirus on chromosome 3 is upregulated in amyotrophic lateral sclerosis motor cortex. <i>Scientific Reports</i> , 2021, 11, 14283.	1.6	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.	1.5	12
92	<i>APOE</i> $\epsilon$ 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.	1.8	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.	0.9	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.	1.5	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.	1.8	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.	1.2	8
97	Frequency and methylation status of selected retrotransposition competent L1 loci in amyotrophic lateral sclerosis. <i>Molecular Brain</i> , 2020, 13, 154.	1.3	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.	3.0	4
99	Axonal injury is detected by $\beta$ APP immunohistochemistry in rapid death from head injury following road traffic collision. <i>International Journal of Legal Medicine</i> , 2022, 136, 1321-1339.	1.2	3
100	A pathologically confirmed case of combined amyotrophic lateral sclerosis with <i>C9orf72</i> mutation and multiple system atrophy. <i>Neuropathology</i> , 0, , .	0.7	3
101	Plasma $\tau$ 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.	0.4	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.	0.4	0