

Leonard Petrucelli

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150 papers	13,729 citations	61 h-index	116 g-index
161 ext. papers	16,876 ext. citations	14.1 avg, IF	6.11 L-index

#	Paper	IF	Citations
150	Unconventional translation of C9ORF72 GGGGCC expansion generates insoluble polypeptides specific to c9FTD/ALS. <i>Neuron</i> , 2013 , 77, 639-46	13.9	783
149	RNA toxicity from the ALS/FTD C9ORF72 expansion is mitigated by antisense intervention. <i>Neuron</i> , 2013 , 80, 415-28	13.9	650
148	GGGGCC repeat expansion in C9orf72 compromises nucleocytoplasmic transport. <i>Nature</i> , 2015 , 525, 129-33	50.4	540
147	Targeting RNA foci in iPSC-derived motor neurons from ALS patients with a C9ORF72 repeat expansion. <i>Science Translational Medicine</i> , 2013 , 5, 208ra149	17.5	488
146	Aberrant cleavage of TDP-43 enhances aggregation and cellular toxicity. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009 , 106, 7607-12	11.5	433
145	Lewy bodies and parkinsonism in families with parkin mutations. <i>Annals of Neurology</i> , 2001 , 50, 293-300	9.4	425
144	Antisense transcripts of the expanded C9ORF72 hexanucleotide repeat form nuclear RNA foci and undergo repeat-associated non-ATG translation in c9FTD/ALS. <i>Acta Neuropathologica</i> , 2013 , 126, 829-44	14.3	392
143	Wild-type human TDP-43 expression causes TDP-43 phosphorylation, mitochondrial aggregation, motor deficits, and early mortality in transgenic mice. <i>Journal of Neuroscience</i> , 2010 , 30, 10851-9	6.6	373
142	TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. <i>Neuron</i> , 2017 , 95, 808-816.e9	13.9	341
141	ER-mitochondria associations are regulated by the VAPB-PTIP51 interaction and are disrupted by ALS/FTD-associated TDP-43. <i>Nature Communications</i> , 2014 , 5, 3996	17.4	341
140	Novel mutations in TARDBP (TDP-43) in patients with familial amyotrophic lateral sclerosis. <i>PLoS Genetics</i> , 2008 , 4, e1000193	6	339
139	Gain of Toxicity from ALS/FTD-Linked Repeat Expansions in C9ORF72 Is Alleviated by Antisense Oligonucleotides Targeting GGGGCC-Containing RNAs. <i>Neuron</i> , 2016 , 90, 535-50	13.9	331
138	An autoradiographic evaluation of AV-1451 Tau PET in dementia. <i>Acta Neuropathologica Communications</i> , 2016 , 4, 58	7.3	305
137	Neurodegeneration. C9ORF72 repeat expansions in mice cause TDP-43 pathology, neuronal loss, and behavioral deficits. <i>Science</i> , 2015 , 348, 1151-4	33.3	279
136	TDP-43 pathology disrupts nuclear pore complexes and nucleocytoplasmic transport in ALS/FTD. <i>Nature Neuroscience</i> , 2018 , 21, 228-239	25.5	240
135	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. <i>Nature Neuroscience</i> , 2015 , 18, 1175-82	25.5	235
134	Discovery of a biomarker and lead small molecules to target r(GGGGCC)-associated defects in c9FTD/ALS. <i>Neuron</i> , 2014 , 83, 1043-50	13.9	232

133	Aggregation-prone c9FTD/ALS poly(GA) RAN-translated proteins cause neurotoxicity by inducing ER stress. <i>Acta Neuropathologica</i> , 2014 , 128, 505-24	14.3	227
132	Poly(GR) in C9ORF72-Related ALS/FTD Compromises Mitochondrial Function and Increases Oxidative Stress and DNA Damage in iPSC-Derived Motor Neurons. <i>Neuron</i> , 2016 , 92, 383-391	13.9	220
131	Reduced C9orf72 gene expression in c9FTD/ALS is caused by histone trimethylation, an epigenetic event detectable in blood. <i>Acta Neuropathologica</i> , 2013 , 126, 895-905	14.3	217
130	C9orf72 BAC Transgenic Mice Display Typical Pathologic Features of ALS/FTD. <i>Neuron</i> , 2015 , 88, 892-901	13.9	201
129	C9ORF72 poly(GA) aggregates sequester and impair HR23 and nucleocytoplasmic transport proteins. <i>Nature Neuroscience</i> , 2016 , 19, 668-677	25.5	201
128	Association between repeat sizes and clinical and pathological characteristics in carriers of C9ORF72 repeat expansions (Xpansize-72): a cross-sectional cohort study. <i>Lancet Neurology</i> , 2013 , 12, 978-88	24.1	200
127	Human C9ORF72 Hexanucleotide Expansion Reproduces RNA Foci and Dipeptide Repeat Proteins but Not Neurodegeneration in BAC Transgenic Mice. <i>Neuron</i> , 2015 , 88, 902-909	13.9	183
126	Converging pathways in neurodegeneration, from genetics to mechanisms. <i>Nature Neuroscience</i> , 2018 , 21, 1300-1309	25.5	183
125	Interaction of tau with the RNA-Binding Protein TIA1 Regulates tau Pathophysiology and Toxicity. <i>Cell Reports</i> , 2016 , 15, 1455-1466	10.6	176
124	Tau Protein Disrupts Nucleocytoplasmic Transport in Alzheimer's Disease. <i>Neuron</i> , 2018 , 99, 925-940.e7	13.9	169
123	Updated TDP-43 in Alzheimer's disease staging scheme. <i>Acta Neuropathologica</i> , 2016 , 131, 571-85	14.3	168
122	Posttranslational Modifications Mediate the Structural Diversity of Tauopathy Strains. <i>Cell</i> , 2020 , 180, 633-644.e12	56.2	156
121	Quantitative analysis and clinico-pathological correlations of different dipeptide repeat protein pathologies in C9ORF72 mutation carriers. <i>Acta Neuropathologica</i> , 2015 , 130, 845-61	14.3	155
120	Differential Toxicity of Nuclear RNA Foci versus Dipeptide Repeat Proteins in a Drosophila Model of C9ORF72 FTD/ALS. <i>Neuron</i> , 2015 , 87, 1207-1214	13.9	149
119	Poly(GR) impairs protein translation and stress granule dynamics in C9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis. <i>Nature Medicine</i> , 2018 , 24, 1136-1142	50.5	149
118	Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , 2017 , 9,	17.5	128
117	Alterations in microRNA-124 and AMPA receptors contribute to social behavioral deficits in frontotemporal dementia. <i>Nature Medicine</i> , 2014 , 20, 1444-51	50.5	125
116	Homozygosity for the C9orf72 GGGGCC repeat expansion in frontotemporal dementia. <i>Acta Neuropathologica</i> , 2013 , 126, 401-9	14.3	119

115	The dual functions of the extreme N-terminus of TDP-43 in regulating its biological activity and inclusion formation. <i>Human Molecular Genetics</i> , 2013 , 22, 3112-22	5.6	119
114	Mechanisms of toxicity in C9FTLD/ALS. <i>Acta Neuropathologica</i> , 2014 , 127, 359-76	14.3	114
113	Heterochromatin anomalies and double-stranded RNA accumulation underlie poly(PR) toxicity. <i>Science</i> , 2019 , 363,	33.3	104
112	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. <i>Science</i> , 2016 , 353, 708-12	33.3	92
111	Tau aggregation influences cognition and hippocampal atrophy in the absence of beta-amyloid: a clinico-imaging-pathological study of primary age-related tauopathy (PART). <i>Acta Neuropathologica</i> , 2017 , 133, 705-715	14.3	91
110	Timing and significance of pathological features in C9orf72 expansion-associated frontotemporal dementia. <i>Brain</i> , 2016 , 139, 3202-3216	11.2	90
109	Microglial translational profiling reveals a convergent APOE pathway from aging, amyloid, and tau. <i>Journal of Experimental Medicine</i> , 2018 , 215, 2235-2245	16.6	85
108	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. <i>Acta Neuropathologica</i> , 2015 , 130, 863-76	14.3	81
107	Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. <i>Nature Neuroscience</i> , 2020 , 23, 615-624	25.5	80
106	An acetylation-phosphorylation switch that regulates tau aggregation propensity and function. <i>Journal of Biological Chemistry</i> , 2017 , 292, 15277-15286	5.4	78
105	CUG initiation and frameshifting enable production of dipeptide repeat proteins from ALS/FTD C9ORF72 transcripts. <i>Nature Communications</i> , 2018 , 9, 152	17.4	77
104	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. <i>Neurology</i> , 2013 , 81, 1332-41	6.5	75
103	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. <i>Acta Neuropathologica</i> , 2015 , 130, 559-73	14.3	72
102	Spinal poly-GA inclusions in a C9orf72 mouse model trigger motor deficits and inflammation without neuron loss. <i>Acta Neuropathologica</i> , 2017 , 134, 241-254	14.3	70
101	Systematic analysis of dark and camouflaged genes reveals disease-relevant genes hiding in plain sight. <i>Genome Biology</i> , 2019 , 20, 97	18.3	68
100	APOE ϵ is associated with increased tau pathology in primary tauopathy. <i>Nature Communications</i> , 2018 , 9, 4388	17.4	68
99	A zebrafish model for C9orf72 ALS reveals RNA toxicity as a pathogenic mechanism. <i>Acta Neuropathologica</i> , 2018 , 135, 427-443	14.3	66
98	Long-read sequencing across the C9orf72 'GGGGCC' repeat expansion: implications for clinical use and genetic discovery efforts in human disease. <i>Molecular Neurodegeneration</i> , 2018 , 13, 46	19	66

97	Loss of clusterin shifts amyloid deposition to the cerebrovasculature via disruption of perivascular drainage pathways. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017 , 114, E6962-E6971	11.5	66
96	Aberrant deposition of stress granule-resident proteins linked to C9orf72-associated TDP-43 proteinopathy. <i>Molecular Neurodegeneration</i> , 2019 , 14, 9	19	64
95	Genetic Convergence Brings Clarity to the Enigmatic Red Line in ALS. <i>Neuron</i> , 2019 , 101, 1057-1069	13.9	63
94	Characterization of DNA hypermethylation in the cerebellum of c9FTD/ALS patients. <i>Brain Research</i> , 2014 , 1584, 15-21	3.7	63
93	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. <i>Human Molecular Genetics</i> , 2017 , 26, 3421-3431	5.6	63
92	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. <i>Neurobiology of Aging</i> , 2014 , 35, 2421.e13-7	5.6	62
91	Misregulation of human sortilin splicing leads to the generation of a nonfunctional progranulin receptor. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012 , 109, 21510-5	11.5	61
90	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> , 2018 , 17, 548-558	24.1	60
89	Phosphorylated neurofilament heavy chain: A biomarker of survival for C9ORF72-associated amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2017 , 82, 139-146	9.4	58
88	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. <i>Acta Neuropathologica</i> , 2017 , 134, 255-269	14.3	57
87	Monitoring peripheral nerve degeneration in ALS by label-free stimulated Raman scattering imaging. <i>Nature Communications</i> , 2016 , 7, 13283	17.4	56
86	The lysosomal protein cathepsin L is a progranulin protease. <i>Molecular Neurodegeneration</i> , 2017 , 12, 55	19	54
85	RPS25 is required for efficient RAN translation of C9orf72 and other neurodegenerative disease-associated nucleotide repeats. <i>Nature Neuroscience</i> , 2019 , 22, 1383-1388	25.5	54
84	Insights into the pathogenic mechanisms of Chromosome 9 open reading frame 72 (C9orf72) repeat expansions. <i>Journal of Neurochemistry</i> , 2016 , 138 Suppl 1, 145-62	6	54
83	TDP-43 functions within a network of hnRNP proteins to inhibit the production of a truncated human SORT1 receptor. <i>Human Molecular Genetics</i> , 2016 , 25, 534-45	5.6	52
82	ALS and FTD: an epigenetic perspective. <i>Acta Neuropathologica</i> , 2016 , 132, 487-502	14.3	51
81	Poly-GR dipeptide repeat polymers correlate with neurodegeneration and Clinicopathological subtypes in C9ORF72-related brain disease. <i>Acta Neuropathologica Communications</i> , 2018 , 6, 63	7.3	51
80	Genome-wide analyses as part of the international FTLD-TDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTLD. <i>Acta Neuropathologica</i> , 2019 , 137, 879-899	14.3	50

79	Disease Mechanisms of Repeat Expansions. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2018 , 8,	5.4	49
78	poly(GR) aggregation induces TDP-43 proteinopathy. <i>Science Translational Medicine</i> , 2020 , 12,	17.5	49
77	TDP-1, the <i>Caenorhabditis elegans</i> ortholog of TDP-43, limits the accumulation of double-stranded RNA. <i>EMBO Journal</i> , 2014 , 33, 2947-66	13	44
76	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	5.6	43
75	The Hairpin Form of r(GC) in c9ALS/FTD Is Repeat-Associated Non-ATG Translated and a Target for Bioactive Small Molecules. <i>Cell Chemical Biology</i> , 2019 , 26, 179-190.e12	8.2	43
74	Association of Apolipoprotein E ϵ 4 With Transactive Response DNA-Binding Protein 43. <i>JAMA Neurology</i> , 2018 , 75, 1347-1354	17.2	42
73	TDP-43 mutations causing amyotrophic lateral sclerosis are associated with altered expression of RNA-binding protein hnRNP K and affect the Nrf2 antioxidant pathway. <i>Human Molecular Genetics</i> , 2017 , 26, 1732-1746	5.6	41
72	TIA1 regulates the generation and response to toxic tau oligomers. <i>Acta Neuropathologica</i> , 2019 , 137, 259-277	14.3	39
71	Toxic expanded GGGGCC repeat transcription is mediated by the PAF1 complex in C9orf72-associated FTD. <i>Nature Neuroscience</i> , 2019 , 22, 863-874	25.5	38
70	Severe amygdala dysfunction in a MAPT transgenic mouse model of frontotemporal dementia. <i>Neurobiology of Aging</i> , 2014 , 35, 1769-77	5.6	37
69	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. <i>Journal of Clinical Investigation</i> , 2020 , 130, 6080-6092	15.9	34
68	The influence of tau, amyloid, alpha-synuclein, TDP-43, and vascular pathology in clinically normal elderly individuals. <i>Neurobiology of Aging</i> , 2019 , 77, 26-36	5.6	32
67	Pathological, imaging and genetic characteristics support the existence of distinct TDP-43 types in non-FTLD brains. <i>Acta Neuropathologica</i> , 2019 , 137, 227-238	14.3	32
66	Poly(GP), neurofilament and grey matter deficits in expansion carriers. <i>Annals of Clinical and Translational Neurology</i> , 2018 , 5, 583-597	5.3	29
65	Linking the VPS35 and EIF4G1 pathways in Parkinson's disease. <i>Neuron</i> , 2015 , 85, 1-3	13.9	29
64	Mutant TDP-43 does not impair mitochondrial bioenergetics in vitro and in vivo. <i>Molecular Neurodegeneration</i> , 2017 , 12, 37	19	28
63	Replication of progressive supranuclear palsy genome-wide association study identifies SLCO1A2 and DUSP10 as new susceptibility loci. <i>Molecular Neurodegeneration</i> , 2018 , 13, 37	19	28
62	ADAR2 mislocalization and widespread RNA editing aberrations in C9orf72-mediated ALS/FTD. <i>Acta Neuropathologica</i> , 2019 , 138, 49-65	14.3	27

61	p53 is a central regulator driving neurodegeneration caused by C9orf72 poly(PR). <i>Cell</i> , 2021 , 184, 689-708.e20	36.2	26
60	Identification and characterization of the human parkin gene promoter. <i>Journal of Neurochemistry</i> , 2001 , 78, 1146-52	6	25
59	Chimeric Peptide Species Contribute to Divergent Dipeptide Repeat Pathology in c9ALS/FTD and SCA36. <i>Neuron</i> , 2020 , 107, 292-305.e6	13.9	25
58	Unaffected mosaic case: RNA foci, dipeptide proteins, but upregulated C9orf72 expression. <i>Neurology</i> , 2018 , 90, e323-e331	6.5	24
57	Expanded C9ORF72 hexanucleotide repeat in depressive pseudodementia. <i>JAMA Neurology</i> , 2014 , 71, 775-81	17.2	24
56	Divergent phenotypes in mutant TDP-43 transgenic mice highlight potential confounds in TDP-43 transgenic modeling. <i>PLoS ONE</i> , 2014 , 9, e86513	3.7	23
55	eIF4B and eIF4H mediate GR production from expanded G4C2 in a Drosophila model for C9orf72-associated ALS. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 62	7.3	22
54	Cross-sectional and longitudinal measures of chitinase proteins in amyotrophic lateral sclerosis and expression of CHI3L1 in activated astrocytes. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020 , 91, 350-358	5.5	22
53	TIA1 potentiates tau phase separation and promotes generation of toxic oligomeric tau. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021 , 118,	11.5	22
52	Elevated methylation levels, reduced expression levels, and frequent contractions in a clinical cohort of C9orf72 expansion carriers. <i>Molecular Neurodegeneration</i> , 2020 , 15, 7	19	20
51	Plasma neurofilament light predicts mortality in patients with stroke. <i>Science Translational Medicine</i> , 2020 , 12,	17.5	20
50	Hexanucleotide Repeat Expansions in c9FTD/ALS and SCA36 Confer Selective Patterns of Neurodegeneration InVivo. <i>Cell Reports</i> , 2020 , 31, 107616	10.6	18
49	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 150	7.3	18
48	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. <i>Experimental Neurology</i> , 2016 , 277, 171-177	5.7	16
47	Serum neurofilament light protein correlates with unfavorable clinical outcomes in hospitalized patients with COVID-19. <i>Science Translational Medicine</i> , 2021 , 13,	17.5	16
46	Dipeptide repeat proteins activate a heat shock response found in C9ORF72-ALS/FTLD patients. <i>Acta Neuropathologica Communications</i> , 2018 , 6, 55	7.3	15
45	Toward allele-specific targeting therapy and pharmacodynamic marker for spinocerebellar ataxia type 3. <i>Science Translational Medicine</i> , 2020 , 12,	17.5	15
44	Tau exhibits unique seeding properties in globular glial tauopathy. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 36	7.3	14

43	Understanding biomarkers of neurodegeneration: Novel approaches to detecting tau pathology. <i>Nature Medicine</i> , 2015 , 21, 219-20	50.5	14
42	Loss of Tmem106b is unable to ameliorate frontotemporal dementia-like phenotypes in an AAV mouse model of C9ORF72-repeat induced toxicity. <i>Acta Neuropathologica Communications</i> , 2018 , 6, 42	7.3	14
41	TDP-43 represses cryptic exon inclusion in the FTD-ALS gene UNC13A.. <i>Nature</i> , 2022 ,	50.4	14
40	Mitophagy alterations in Alzheimer's disease are associated with granulovacuolar degeneration and early tau pathology. <i>Alzheimer's and Dementia</i> , 2020 , 17, 417	1.2	13
39	Utility of FDG-PET in diagnosis of Alzheimer-related TDP-43 proteinopathy. <i>Neurology</i> , 2020 , 95, e23-e34	46.5	11
38	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. <i>Annals of Clinical and Translational Neurology</i> , 2019 , 6, 1782-1796	5.3	11
37	FTDP-17 with Pick body-like inclusions associated with a novel tau mutation, p.E372G. <i>Brain Pathology</i> , 2017 , 27, 612-626	6	11
36	Astrocyte-derived clusterin suppresses amyloid formation in vivo. <i>Molecular Neurodegeneration</i> , 2020 , 15, 71	19	11
35	Interaction of tau with HNRNPA2B1 and N-methyladenosine RNA mediates the progression of tauopathy. <i>Molecular Cell</i> , 2021 , 81, 4209-4227.e12	17.6	11
34	Cellular and pathological heterogeneity of primary tauopathies. <i>Molecular Neurodegeneration</i> , 2021 , 16, 57	19	11
33	OPTN p.Met468Arg and ATXN2 intermediate length polyQ extension in families with C9orf72 mediated amyotrophic lateral sclerosis and frontotemporal dementia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018 , 177, 75-85	3.5	10
32	C-terminal and full length TDP-43 specie differ according to FTLT-TDP lesion type but not genetic mutation. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 100	7.3	9
31	Abnormal expression of homeobox genes and transthyretin in expansion carriers. <i>Neurology: Genetics</i> , 2017 , 3, e161	3.8	9
30	Premature termination codon readthrough upregulates progranulin expression and improves lysosomal function in preclinical models of GRN deficiency. <i>Molecular Neurodegeneration</i> , 2020 , 15, 21	19	8
29	The Caenorhabditis elegans Ortholog of TDP-43 Regulates the Chromatin Localization of the Heterochromatin Protein 1 Homolog HPL-2. <i>Molecular and Cellular Biology</i> , 2018 , 38,	4.8	8
28	The AD tau core spontaneously self-assembles and recruits full-length tau to filaments. <i>Cell Reports</i> , 2021 , 34, 108843	10.6	8
27	Ribonuclease recruitment using a small molecule reduced c9ALS/FTD r(GC) repeat expansion in vitro and in vivo ALS models. <i>Science Translational Medicine</i> , 2021 , 13, eabd5991	17.5	6
26	Deep vein thrombosis and pulmonary embolism among hospitalized coronavirus disease 2019-positive patients predicted for higher mortality and prolonged intensive care unit and hospital stays in a multisite healthcare system. <i>Journal of Vascular Surgery: Venous and Lymphatic Disorders</i> , 2021 , 9, 1361-1370.e1	3.2	6

25	Clusterin ameliorates tau pathology in vivo by inhibiting fibril formation. <i>Acta Neuropathologica Communications</i> , 2020 , 8, 210	7.3	5
24	TRIO gene segregation in a family with cerebellar ataxia. <i>Neurologia I Neurochirurgia Polska</i> , 2018 , 52, 743-749	1	5
23	Microglial lysosome dysfunction contributes to white matter pathology and TDP-43 proteinopathy in GRN-associated FTD. <i>Cell Reports</i> , 2021 , 36, 109581	10.6	5
22	Homotypic fibrillization of TMEM106B across diverse neurodegenerative diseases.. <i>Cell</i> , 2022 ,	56.2	5
21	ARHGEF28 p.Lys280Metfs40Ter in an amyotrophic lateral sclerosis family with a C9orf72 expansion. <i>Neurology: Genetics</i> , 2017 , 3, e190	3.8	4
20	Structural Features of Small Molecules Targeting the RNA Repeat Expansion That Causes Genetically Defined ALS/FTD. <i>ACS Chemical Biology</i> , 2020 , 15, 3112-3123	4.9	4
19	Urine levels of the polyglutamine ataxin-3 protein are elevated in patients with spinocerebellar ataxia type 3. <i>Parkinsonism and Related Disorders</i> , 2021 , 89, 151-154	3.6	3
18	Application of a bioinformatic pipeline to RNA-seq data identifies novel virus-like sequence in human blood. <i>G3: Genes, Genomes, Genetics</i> , 2021 , 11,	3.2	2
17	Long-read targeted sequencing uncovers clinicopathological associations for C9orf72-linked diseases. <i>Brain</i> , 2021 , 144, 1082-1088	11.2	2
16	Poly(GR) and poly(GA) in cerebrospinal fluid as potential biomarkers for C9ORF72-ALS/FTD.. <i>Nature Communications</i> , 2022 , 13, 2799	17.4	2
15	Enhanced phosphorylation of T153 in soluble tau is a defining biochemical feature of the A152T tau risk variant. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 10	7.3	1
14	Long-read sequencing across the C9orf72 CAGGCC repeat expansion: implications for clinical use and genetic discovery efforts in human disease		1
13	NIH funding trends for neurosurgeon-scientists from 1993-2017: Biomedical workforce implications for neurooncology. <i>Journal of Neuro-Oncology</i> , 2021 , 154, 51-62	4.8	1
12	Amyotrophic lateral sclerosis - insight into susceptibility.. <i>Nature Reviews Neurology</i> , 2022 ,	15	1
11	Modelling amyotrophic lateral sclerosis in rodents.. <i>Nature Reviews Neuroscience</i> , 2022 ,	13.5	1
10	HDAC6 Interacts With Poly (GA) and Modulates its Accumulation in c9FTD/ALS.. <i>Frontiers in Cell and Developmental Biology</i> , 2021 , 9, 809942	5.7	0
9	TDP-43-associated atrophy in brains with and without frontotemporal lobar degeneration.. <i>NeuroImage: Clinical</i> , 2022 , 34, 102954	5.3	0
8	A Small Molecule Exploits Hidden Structural Features within the RNA Repeat Expansion That Causes c9ALS/FTD and Rescues Pathological Hallmarks. <i>ACS Chemical Neuroscience</i> , 2021 , 12, 4076-4089	5.7	0

7	Plasma PolyQ-ATXN3 Levels Associate With Cerebellar Degeneration and Behavioral Abnormalities in a New AAV-Based SCA3 Mouse Model.. <i>Frontiers in Cell and Developmental Biology</i> , 2022 , 10, 863089	5.7	o
6	Comprehensive cross-sectional and longitudinal analyses of plasma neurofilament light across FTD spectrum disorders.. <i>Cell Reports Medicine</i> , 2022 , 3, 100607	18	o
5	Cracking the cryptic code in amyotrophic lateral sclerosis and frontotemporal dementia: Towards therapeutic targets and biomarkers.. <i>Clinical and Translational Medicine</i> , 2022 , 12, e818	5.7	o
4	AlPuts the Alpha in Synuclein. <i>Neuron</i> , 2020 , 105, 205-206	13.9	
3	Epigenetic modifications of the C9ORF72 gene: a potential biomarker of disease?. <i>Future Neurology</i> , 2014 , 9, 123-126	1.5	
2	O1-07-01: Accelerated lipofuscinosis and ubiquitination in granulin knockout mice suggests a role for progranulin in successful aging 2010 , 6, S83-S83		
1	Comment on: Polyglutamine-Expanded Ataxin-3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood.. <i>Movement Disorders</i> , 2022 , 37, 1120-1121	7	