Leonard Petrucelli

List of Publications by Year
in descending order

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153 papers	19,518 citations	13068 68 h-index	12910 131 g-index
161 all docs	161 docs citations	161 times ranked	15214 citing authors

#	Article	IF	Citations
1	TDP-43-associated atrophy in brains with and without frontotemporal lobar degeneration. NeuroImage: Clinical, 2022, 34, 102954.	1.4	3
2	TDP-43 represses cryptic exon inclusion in the FTDâ€“ALS gene UNC13A. Nature, 2022, 603, 124-130.	13.7	193
3	Amyotrophic lateral sclerosis â€” insight into susceptibility. Nature Reviews Neurology, 2022, 18, 189-190.	4.9	1
4	Homotypic fibrillization of TMEM106B across diverse neurodegenerative diseases. Cell, 2022, 185, 1346-1355.e15.	13.5	70
5	Plasma PolyQ-ATXN3 Levels Associate With Cerebellar Degeneration and Behavioral Abnormalities in a New AAV-Based SCA3 Mouse Model. Frontiers in Cell and Developmental Biology, 2022, 10, 863089.	1.8	5
6	Modelling amyotrophic lateral sclerosis in rodents. Nature Reviews Neuroscience, 2022, 23, 231-251.	4.9	17
7	Shared brain transcriptomic signature in TDP-43 type A FTLD patients with or without GRN mutations. Brain, 2022, 145, 2472-2485.	3.7	6
8	Comprehensive cross-sectional and longitudinal analyses of plasma neurofilament light across FTD spectrum disorders. Cell Reports Medicine, 2022, 3, 100607.	3.3	21
9	Tau and neurofilament lightâ€chain as fluid biomarkers in spinocerebellar ataxia type 3. European Journal of Neurology, 2022, 29, 2439-2452.	1.7	25
10	Cracking the cryptic code in amyotrophic lateral sclerosis and frontotemporal dementia: Towards therapeutic targets and biomarkers. Clinical and Translational Medicine, 2022, 12, e818.	1.7	11
11	Comment on: <scp>Polyglutamineâ€Expanded</scp> Ataxinâ€3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood. Movement Disorders, 2022, 37, 1120-1121.	2.2	0
12	Poly(GR) and poly(GA) in cerebrospinal fluid as potential biomarkers for C9ORF72-ALS/FTD. Nature Communications, 2022, 13, 2799.	5.8	28
13	Mitophagy alterations in Alzheimer's disease are associated with granulovacuolar degeneration and early tau pathology. Alzheimer's and Dementia, 2021, 17, 417-430.	0.4	34
14	p53 is a central regulator driving neurodegeneration caused by C9orf72 poly(PR). Cell, 2021, 184, 689-708.e20.	13.5	104
15	TIA1 potentiates tau phase separation and promotes generation of toxic oligomeric tau. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	72
16	The AD tau core spontaneously self-assembles and recruits full-length tau to filaments. Cell Reports, 2021, 34, 108843.	2.9	30
17	Application of a bioinformatic pipeline to RNA-seq data identifies novel virus-like sequence in human blood. G3: Genes, Genomes, Genetics, 2021, 11, .	0.8	4
18	Deep vein thrombosis and pulmonary embolism among hospitalized coronavirus disease 2019â€“positive patients predicted for higher mortality and prolonged intensive care unit and hospital stays in a multisite healthcare system. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2021, 9, 1361-1370.e1.	0.9	17

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19	Long-read targeted sequencing uncovers clinicopathological associations for C9orf72-linked diseases. Brain, 2021, 144, 1082-1088.	3.7	17
20	Serum neurofilament light protein correlates with unfavorable clinical outcomes in hospitalized patients with COVID-19. Science Translational Medicine, 2021, 13, .	5.8	67
21	NIH funding trends for neurosurgeon-scientists from 1993â€“2017: Biomedical workforce implications for neurooncology. Journal of Neuro-Oncology, 2021, 154, 51-62.	1.4	6
22	Urine levels of the polyglutamine ataxin-3 protein are elevated in patients with spinocerebellar ataxia type 3. Parkinsonism and Related Disorders, 2021, 89, 151-154.	1.1	9
23	Interaction of tau with HNRNPA2B1 and N6-methyladenosine RNA mediates the progression of tauopathy. Molecular Cell, 2021, 81, 4209-4227.e12.	4.5	84
24	Cellular and pathological heterogeneity of primary tauopathies. Molecular Neurodegeneration, 2021, 16, 57.	4.4	85
25	Microglial lysosome dysfunction contributes to white matter pathology and TDP-43 proteinopathy in GRN-associated FTD. Cell Reports, 2021, 36, 109581.	2.9	33
26	Ribonuclease recruitment using a small molecule reduced c9ALS/FTD r(G ₄ C ₂) Tj ETQq0 0 0 rgBT /Overlock 1	5.8	39
27	A Small Molecule Exploits Hidden Structural Features within the RNA Repeat Expansion That Causes c9ALS/FTD and Rescues Pathological Hallmarks. ACS Chemical Neuroscience, 2021, 12, 4076-4089.	1.7	8
28	HDAC6 Interacts With Poly (GA) and Modulates its Accumulation in c9FTD/ALS. Frontiers in Cell and Developmental Biology, 2021, 9, 809942.	1.8	4
29	Toward allele-specific targeting therapy and pharmacodynamic marker for spinocerebellar ataxia type 3. Science Translational Medicine, 2020, 12, .	5.8	32
30	Astrocyte-derived clusterin suppresses amyloid formation in vivo. Molecular Neurodegeneration, 2020, 15, 71.	4.4	26
31	Plasma neurofilament light predicts mortality in patients with stroke. Science Translational Medicine, 2020, 12, .	5.8	51
32	Clusterin ameliorates tau pathology in vivo by inhibiting fibril formation. Acta Neuropathologica Communications, 2020, 8, 210.	2.4	24
33	Structural Features of Small Molecules Targeting the RNA Repeat Expansion That Causes Genetically Defined ALS/FTD. ACS Chemical Biology, 2020, 15, 3112-3123.	1.6	12
34	C9orf72 poly(GR) aggregation induces TDP-43 proteinopathy. Science Translational Medicine, 2020, 12, .	5.8	115
35	Cross-sectional and longitudinal measures of chitinase proteins in amyotrophic lateral sclerosis and expression of CHI3L1 in activated astrocytes. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 350-358.	0.9	54
36	Hexanucleotide Repeat Expansions in c9FTD/ALS and SCA36 Confer Selective Patterns of Neurodegeneration InÂVivo. Cell Reports, 2020, 31, 107616.	2.9	37

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37	Utility of FDG-PET in diagnosis of Alzheimer-related TDP-43 proteinopathy. Neurology, 2020, 95, e23-e34.	1.5	27
38	Premature termination codon readthrough upregulates progranulin expression and improves lysosomal function in preclinical models of GRN deficiency. Molecular Neurodegeneration, 2020, 15, 21.	4.4	19
39	AÎ² Puts the Alpha in Synuclein. Neuron, 2020, 105, 205-206.	3.8	2
40	Elevated methylation levels, reduced expression levels, and frequent contractions in a clinical cohort of C9orf72 expansion carriers. Molecular Neurodegeneration, 2020, 15, 7.	4.4	34
41	Posttranslational Modifications Mediate the Structural Diversity of Tauopathy Strains. Cell, 2020, 180, 633-644.e12.	13.5	300
42	Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. Nature Neuroscience, 2020, 23, 615-624.	7.1	157
43	Chimeric Peptide Species Contribute to Divergent Dipeptide Repeat Pathology in c9ALS/FTD and SCA36. Neuron, 2020, 107, 292-305.e6.	3.8	51
44	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	3.9	117
45	RPS25 is required for efficient RAN translation of C9orf72 and other neurodegenerative disease-associated nucleotide repeats. Nature Neuroscience, 2019, 22, 1383-1388.	7.1	87
46	C-terminal and full length TDP-43 specie differ according to FTLD-TDP lesion type but not genetic mutation. Acta Neuropathologica Communications, 2019, 7, 100.	2.4	11
47	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. Acta Neuropathologica Communications, 2019, 7, 150.	2.4	40
48	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 1782-1796.	1.7	20
49	The influence of tau, amyloid, alpha-synuclein, TDP-43, and vascular pathology in clinically normal elderly individuals. Neurobiology of Aging, 2019, 77, 26-36.	1.5	51
50	Enhanced phosphorylation of T153 in soluble tau is a defining biochemical feature of the A152T tau risk variant. Acta Neuropathologica Communications, 2019, 7, 10.	2.4	3
51	Toxic expanded GGGGCC repeat transcription is mediated by the PAF1 complex in C9orf72-associated FTD. Nature Neuroscience, 2019, 22, 863-874.	7.1	65
52	Systematic analysis of dark and camouflaged genes reveals disease-relevant genes hiding in plain sight. Genome Biology, 2019, 20, 97.	3.8	122
53	eIF4B and eIF4H mediate GR production from expanded G4C2 in a Drosophila model for C9orf72-associated ALS. Acta Neuropathologica Communications, 2019, 7, 62.	2.4	38
54	Genetic Convergence Brings Clarity to the Enigmatic Red Line in ALS. Neuron, 2019, 101, 1057-1069.	3.8	111

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55	Tau exhibits unique seeding properties in globular glial tauopathy. Acta Neuropathologica Communications, 2019, 7, 36.	2.4	28
56	ADAR2 mislocalization and widespread RNA editing aberrations in C9orf72-mediated ALS/FTD. Acta Neuropathologica, 2019, 138, 49-65.	3.9	48
57	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. Acta Neuropathologica, 2019, 137, 879-899.	3.9	90
58	Aberrant deposition of stress granule-resident proteins linked to C9orf72-associated TDP-43 proteinopathy. Molecular Neurodegeneration, 2019, 14, 9.	4.4	111
59	Heterochromatin anomalies and double-stranded RNA accumulation underlie C9orf72 poly(PR) toxicity. Science, 2019, 363, .	6.0	181
60	Pathological, imaging and genetic characteristics support the existence of distinct TDP-43 types in non-FTLD brains. Acta Neuropathologica, 2019, 137, 227-238.	3.9	65
61	The Hairpin Form of r(G4C2)exp in c9ALS/FTD Is Repeat-Associated Non-ATG Translated and a Target for Bioactive Small Molecules. Cell Chemical Biology, 2019, 26, 179-190.e12.	2.5	80
62	TIA1 regulates the generation and response to toxic tau oligomers. Acta Neuropathologica, 2019, 137, 259-277.	3.9	74
63	Poly(GP), neurofilament and grey matter deficits in C9orf72 expansion carriers. Annals of Clinical and Translational Neurology, 2018, 5, 583-597.	1.7	48
64	Unaffected mosaic C9orf72 case. Neurology, 2018, 90, e323-e331.	1.5	33
65	CUG initiation and frameshifting enable production of dipeptide repeat proteins from ALS/FTD C9ORF72 transcripts. Nature Communications, 2018, 9, 152.	5.8	123
66	TDP-43 pathology disrupts nuclear pore complexes and nucleocytoplasmic transport in ALS/FTD. Nature Neuroscience, 2018, 21, 228-239.	7.1	404
67	A zebrafish model for C9orf72 ALS reveals RNA toxicity as a pathogenic mechanism. Acta Neuropathologica, 2018, 135, 427-443.	3.9	98
68	CRISPR expands insight into the mechanisms of ALS and FTD. Nature Reviews Neurology, 2018, 14, 321-323.	4.9	3
69	Cover Image, Volume 177B, Number 1, January 2018. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, i.	1.1	0
70	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	4.9	97
71	Disease Mechanisms ofC9ORF72Repeat Expansions. Cold Spring Harbor Perspectives in Medicine, 2018, 8, a024224.	2.9	75
72	OPTN p.Met468Arg and ATXN2 intermediate length polyQ extension in families with C9orf72 mediated amyotrophic lateral sclerosis and frontotemporal dementia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 75-85.	1.1	12

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73	Converging pathways in neurodegeneration, from genetics to mechanisms. Nature Neuroscience, 2018, 21, 1300-1309.	7.1	325
74	Association of Apolipoprotein E Îµ4 With Transactive Response DNA-Binding Protein 43. JAMA Neurology, 2018, 75, 1347.	4.5	60
75	APOE Îµ2 is associated with increased tau pathology in primary tauopathy. Nature Communications, 2018, 9, 4388.	5.8	100
76	TRIO gene segregation in a family with cerebellar ataxia. Neurologia I Neurochirurgia Polska, 2018, 52, 743-749.	0.6	5
77	Tau Protein Disrupts Nucleocytoplasmic Transport in Alzheimerâ€™s Disease. Neuron, 2018, 99, 925-940.e7.	3.8	302
78	Poly(GR) impairs protein translation and stress granule dynamics in C9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis. Nature Medicine, 2018, 24, 1136-1142.	15.2	241
79	Loss of Tmem106b is unable to ameliorate frontotemporal dementia-like phenotypes in an AAV mouse model of C9ORF72-repeat induced toxicity. Acta Neuropathologica Communications, 2018, 6, 42.	2.4	20
80	Replication of progressive supranuclear palsy genome-wide association study identifies SLCO1A2 and DUSP10 as new susceptibility loci. Molecular Neurodegeneration, 2018, 13, 37.	4.4	54
81	Microglial translational profiling reveals a convergent APOE pathway from aging, amyloid, and tau. Journal of Experimental Medicine, 2018, 215, 2235-2245.	4.2	167
82	Poly-GR dipeptide repeat polymers correlate with neurodegeneration and Clinicopathological subtypes in C9ORF72-related brain disease. Acta Neuropathologica Communications, 2018, 6, 63.	2.4	79
83	Dipeptide repeat proteins activate a heat shock response found in C9ORF72-ALS/FTLD patients. Acta Neuropathologica Communications, 2018, 6, 55.	2.4	24
84	Long-read sequencing across the C9orf72 â€˜GGGGCCâ€™ repeat expansion: implications for clinical use and genetic discovery efforts in human disease. Molecular Neurodegeneration, 2018, 13, 46.	4.4	111
85	The Caenorhabditis elegans Ortholog of TDP-43 Regulates the Chromatin Localization of the Heterochromatin Protein 1 Homolog HPL-2. Molecular and Cellular Biology, 2018, 38, .	1.1	14
86	Tau aggregation influences cognition and hippocampal atrophy in the absence of beta-amyloid: a clinico-imaging-pathological study of primary age-related tauopathy (PART). Acta Neuropathologica, 2017, 133, 705-715.	3.9	125
87	Spinal poly-GA inclusions in a C9orf72 mouse model trigger motor deficits and inflammation without neuron loss. Acta Neuropathologica, 2017, 134, 241-254.	3.9	99
88	Mutant TDP-43 does not impair mitochondrial bioenergetics in vitro and in vivo. Molecular Neurodegeneration, 2017, 12, 37.	4.4	37
89	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. Acta Neuropathologica, 2017, 134, 255-269.	3.9	76
90	Phosphorylated neurofilament heavy chain: A biomarker of survival for <scp>C9ORF</scp>72â€associated amyotrophic lateral sclerosis. Annals of Neurology, 2017, 82, 139-146.	2.8	88

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91	TDP-43 mutations causing amyotrophic lateral sclerosis are associated with altered expression of RNA-binding protein hnRNP K and affect the Nrf2 antioxidant pathway. Human Molecular Genetics, 2017, 26, 1732-1746.	1.4	62
92	Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72 -associated amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	179
93	C9orf72 poly GA RAN-translated protein plays a key role in amyotrophic lateral sclerosis via aggregation and toxicity. Human Molecular Genetics, 2017, 26, 4765-4777.	1.4	64
94	ARHGEF28 p.Lys280Metfs40Ter in an amyotrophic lateral sclerosis family with a C9orf72 expansion. Neurology: Genetics, 2017, 3, e190.	0.9	6
95	An acetylationâ€“phosphorylation switch that regulates tau aggregation propensity and function. Journal of Biological Chemistry, 2017, 292, 15277-15286.	1.6	100
96	TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. Neuron, 2017, 95, 808-816.e9.	3.8	493
97	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. Human Molecular Genetics, 2017, 26, 3421-3431.	1.4	101
98	Loss of clusterin shifts amyloid deposition to the cerebrovasculature via disruption of perivascular drainage pathways. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, E6962-E6971.	3.3	96
99	Abnormal expression of homeobox genes and transthyretin in C9ORF72 expansion carriers. Neurology: Genetics, 2017, 3, e161.	0.9	12
100	FTDPâ€17 with Pick bodyâ€like inclusions associated with a novel tau mutation, p.E372G. Brain Pathology, 2017, 27, 612-626.	2.1	11
101	The lysosomal protein cathepsin L is a progranulin protease. Molecular Neurodegeneration, 2017, 12, 55.	4.4	81
102	Gain of Toxicity from ALS/FTD-Linked Repeat Expansions in C9ORF72 Is Alleviated by Antisense Oligonucleotides Targeting GGGGCC-Containing RNAs. Neuron, 2016, 90, 535-550.	3.8	437
103	Poly(GR) in C9ORF72 -Related ALS/FTD Compromises Mitochondrial Function and Increases Oxidative Stress and DNA Damage in iPSC-Derived Motor Neurons. Neuron, 2016, 92, 383-391.	3.8	323
104	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. Science, 2016, 353, 708-712.	6.0	116
105	Insights into the pathogenic mechanisms of Chromosome 9 open reading frame 72 (C9orf72) repeat expansions. Journal of Neurochemistry, 2016, 138, 145-162.	2.1	59
106	An autoradiographic evaluation of AV-1451 Tau PET in dementia. Acta Neuropathologica Communications, 2016, 4, 58.	2.4	388
107	Timing and significance of pathological features inC9orf72expansion-associated frontotemporal dementia. Brain, 2016, 139, 3202-3216.	3.7	136
108	Monitoring peripheral nerve degeneration in ALS by label-free stimulated Raman scattering imaging. Nature Communications, 2016, 7, 13283.	5.8	82

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109	ALS and FTD: an epigenetic perspective. Acta Neuropathologica, 2016, 132, 487-502.	3.9	60
110	Interaction of tau with the RNA-Binding Protein TIA1 Regulates tau Pathophysiology and Toxicity. Cell Reports, 2016, 15, 1455-1466.	2.9	260
111	Updated TDP-43 in Alzheimerâ€™s disease staging scheme. Acta Neuropathologica, 2016, 131, 571-585.	3.9	244
112	TDP-43 functions within a network of hnRNP proteins to inhibit the production of a truncated human SORT1 receptor. Human Molecular Genetics, 2016, 25, 534-545.	1.4	70
113	C9ORF72 poly(GA) aggregates sequester and impair HR23 and nucleocytoplasmic transport proteins. Nature Neuroscience, 2016, 19, 668-677.	7.1	268
114	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. Experimental Neurology, 2016, 277, 171-177.	2.0	21
115	Linking the VPS35 and EIF4G1 Pathways in Parkinsonâ€™s Disease. Neuron, 2015, 85, 1-3.	3.8	33
116	Understanding Biomarkers of Neurodegeneration: Novel approaches to detecting tau pathology. Nature Medicine, 2015, 21, 219-220.	15.2	15
117	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. Nature Neuroscience, 2015, 18, 1175-1182.	7.1	330
118	C9ORF72 repeat expansions in mice cause TDP-43 pathology, neuronal loss, and behavioral deficits. Science, 2015, 348, 1151-1154.	6.0	332
119	Differential Toxicity of Nuclear RNA Foci versus Dipeptide Repeat Proteins in a Drosophila Model of C9ORF72 FTD/ALS. Neuron, 2015, 87, 1207-1214.	3.8	176
120	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. Acta Neuropathologica, 2015, 130, 863-876.	3.9	104
121	GGGGCC repeat expansion in C9orf72 compromises nucleocytoplasmic transport. Nature, 2015, 525, 129-133.	13.7	692
122	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. Acta Neuropathologica, 2015, 130, 559-573.	3.9	89
123	Quantitative analysis and clinico-pathological correlations of different dipeptide repeat protein pathologies in C9ORF72 mutation carriers. Acta Neuropathologica, 2015, 130, 845-861.	3.9	204
124	C9orf72 BAC Transgenic Mice Display Typical Pathologic Features of ALS/FTD. Neuron, 2015, 88, 892-901.	3.8	249
125	Human C9ORF72 Hexanucleotide Expansion Reproduces RNA Foci and Dipeptide Repeat Proteins but Not Neurodegeneration in BAC Transgenic Mice. Neuron, 2015, 88, 902-909.	3.8	219
126	ERâ€“mitochondria associations are regulated by the VAPBâ€“PTPIP51 interaction and are disrupted by ALS/FTD-associated TDP-43. Nature Communications, 2014, 5, 3996.	5.8	463

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127	Alterations in microRNA-124 and AMPA receptors contribute to social behavioral deficits in frontotemporal dementia. Nature Medicine, 2014, 20, 1444-1451.	15.2	165
128	<scp>TDP</scp>â€1, the <scp>C</scp>aenorhabditis elegans ortholog of <scp>TDP</scp>â€43, limits the accumulation of doubleâ€stranded <scp>RNA</scp>. EMBO Journal, 2014, 33, 2947-2966.	3.5	62
129	Aggregation-prone c9FTD/ALS poly(GA) RAN-translated proteins cause neurotoxicity by inducing ER stress. Acta Neuropathologica, 2014, 128, 505-524.	3.9	284
130	ExpandedC9ORF72Hexanucleotide Repeat in Depressive Pseudodementia. JAMA Neurology, 2014, 71, 775.	4.5	28
131	Mechanisms of toxicity in C9FTLD/ALS. Acta Neuropathologica, 2014, 127, 359-376.	3.9	134
132	Characterization of DNA hypermethylation in the cerebellum of c9FTD/ALS patients. Brain Research, 2014, 1584, 15-21.	1.1	70
133	Severe amygdala dysfunction in a MAPT transgenic mouse model of frontotemporal dementia. Neurobiology of Aging, 2014, 35, 1769-1777.	1.5	48
134	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. Neurobiology of Aging, 2014, 35, 2421.e13-2421.e17.	1.5	74
135	Discovery of a Biomarker and Lead Small Molecules to Target r(GGGGCC)-Associated Defects in c9FTD/ALS. Neuron, 2014, 83, 1043-1050.	3.8	289
136	Epigenetic modifications of theC9ORF72gene: a potential biomarker of disease?. Future Neurology, 2014, 9, 123-126.	0.9	0
137	Divergent Phenotypes in Mutant TDP-43 Transgenic Mice Highlight Potential Confounds in TDP-43 Transgenic Modeling. PLoS ONE, 2014, 9, e86513.	1.1	23
138	Homozygosity for the C9orf72 GGGGCC repeat expansion in frontotemporal dementia. Acta Neuropathologica, 2013, 126, 401-409.	3.9	126
139	Targeting RNA Foci in iPSC-Derived Motor Neurons from ALS Patients with a C9ORF72 Repeat Expansion. Science Translational Medicine, 2013, 5, 208ra149.	5.8	586
140	Association between repeat sizes and clinical and pathological characteristics in carriers of C9ORF72 repeat expansions (Xpansize-72): a cross-sectional cohort study. Lancet Neurology, The, 2013, 12, 978-988.	4.9	232
141	Unconventional Translation of C9ORF72 GGGGCC Expansion Generates Insoluble Polypeptides Specific to c9FTD/ALS. Neuron, 2013, 77, 639-646.	3.8	962
142	RNA Toxicity from the ALS/FTD C9ORF72 Expansion Is Mitigated by Antisense Intervention. Neuron, 2013, 80, 415-428.	3.8	785
143	Antisense transcripts of the expanded C9ORF72 hexanucleotide repeat form nuclear RNA foci and undergo repeat-associated non-ATG translation in c9FTD/ALS. Acta Neuropathologica, 2013, 126, 829-844.	3.9	506
144	Reduced C9orf72 gene expression in c9FTD/ALS is caused by histone trimethylation, an epigenetic event detectable in blood. Acta Neuropathologica, 2013, 126, 895-905.	3.9	263

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145	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. Neurology, 2013, 81, 1332-1341.	1.5	84
146	The dual functions of the extreme N-terminus of TDP-43 in regulating its biological activity and inclusion formation. Human Molecular Genetics, 2013, 22, 3112-3122.	1.4	156
147	Misregulation of human sortilin splicing leads to the generation of a nonfunctional progranulin receptor. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 21510-21515.	3.3	82
148	Wild-Type Human TDP-43 Expression Causes TDP-43 Phosphorylation, Mitochondrial Aggregation, Motor Deficits, and Early Mortality in Transgenic Mice. Journal of Neuroscience, 2010, 30, 10851-10859.	1.7	457
149	O1-07-01: Accelerated lipofuscinosis and ubiquitination in granulin knockout mice suggests a role for progranulin in successful aging. , 2010, 6, S83-S83.		0
150	Aberrant cleavage of TDP-43 enhances aggregation and cellular toxicity. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 7607-7612.	3.3	523
151	Novel Mutations in TARDBP (TDP-43) in Patients with Familial Amyotrophic Lateral Sclerosis. PLoS Genetics, 2008, 4, e1000193.	1.5	393
152	Identification and characterization of the human parkin gene promoter. Journal of Neurochemistry, 2001, 78, 1146-1152.	2.1	31
153	Lewy bodies and parkinsonism in families withparkin mutations. Annals of Neurology, 2001, 50, 293-300.	2.8	479