Leonard Petrucelli

List of Publications by Year in Descending Order

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	TDP-43-associated atrophy in brains with and without frontotemporal lobar degeneration.. *NeuroImage: Clinical*, **2022**, 34, 102954	5.3	0
149	TDP-43 represses cryptic exon inclusion in the FTD-ALS gene UNC13A.. *Nature*, **2022**,	50.4	14
148	Amyotrophic lateral sclerosis - insight into susceptibility.. *Nature Reviews Neurology*, **2022**,	15	1
147	Homotypic fibrillization of TMEM106B across diverse neurodegenerative diseases.. *Cell*, **2022**,	56.2	5
146	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	5.7	0
145	Modelling amyotrophic lateral sclerosis in rodents.. *Nature Reviews Neuroscience*, **2022**,	13.5	1
144	Comprehensive cross-sectional and longitudinal analyses of plasma neurofilament light across FTD spectrum disorders.. *Cell Reports Medicine*, **2022**, 3, 100607	18	0
143	Cracking the cryptic code in amyotrophic lateral sclerosis and frontotemporal dementia: Towards therapeutic targets and biomarkers.. *Clinical and Translational Medicine*, **2022**, 12, e818	5.7	0
142	Comment on: Polyglutamine-Expanded Ataxin-3: A Target Engagement Marker for Spinocerebellar Ataxia Type 3 in Peripheral Blood.. *Movement Disorders*, **2022**, 37, 1120-1121	7	
141	Poly(GR) and poly(GA) in cerebrospinal fluid as potential biomarkers for C9ORF72-ALS/FTD.. *Nature Communications*, **2022**, 13, 2799	17.4	2
140	HDAC6 Interacts With Poly (GA) and Modulates its Accumulation in c9FTD/ALS.. *Frontiers in Cell and Developmental Biology*, **2021**, 9, 809942	5.7	0
139	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	5.7	0
138	Ribonuclease recruitment using a small molecule reduced c9ALS/FTD r(GC) repeat expansion in vitro and in vivo ALS models. *Science Translational Medicine*, **2021**, 13, eabd5991	17.5	6
137	Application of a bioinformatic pipeline to RNA-seq data identifies novel virus-like sequence in human blood. *G3: Genes, Genomes, Genetics*, **2021**, 11,	3.2	2
136	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. *Journal of Vascular Surgery: Venous and Lymphatic Disorders*, **2021**, 9, 1361-1370.e1	3.2	6
135	Long-read targeted sequencing uncovers clinicopathological associations for C9orf72-linked diseases. *Brain*, **2021**, 144, 1082-1088	11.2	2
134	Serum neurofilament light protein correlates with unfavorable clinical outcomes in hospitalized patients with COVID-19. *Science Translational Medicine*, **2021**, 13,	17.5	16

133	p53 is a central regulator driving neurodegeneration caused by C9orf72 poly(PR). *Cell*, **2021**, 184, 689-708.e20	56.2	26
132	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,	11.5	22
131	The AD tau core spontaneously self-assembles and recruits full-length tau to filaments. *Cell Reports* , **2021**, 34, 108843	10.6	8
130	NIH funding trends for neurosurgeon-scientists from 1993-2017: Biomedical workforce implications for neurooncology. *Journal of Neuro-Oncology*, **2021**, 154, 51-62	4.8	1
129	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	3.6	3
128	Interaction of tau with HNRNPA2B1 and N-methyladenosine RNA mediates the progression of tauopathy. *Molecular Cell*, **2021**, 81, 4209-4227.e12	17.6	11
127	Cellular and pathological heterogeneity of primary tauopathies. *Molecular Neurodegeneration*, **2021** , 16, 57	19	11
126	Microglial lysosome dysfunction contributes to white matter pathology and TDP-43 proteinopathy in GRN-associated FTD. *Cell Reports*, **2021**, 36, 109581	10.6	5
125	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	5.5	22
124	Hexanucleotide Repeat Expansions in c9FTD/ALS and SCA36 Confer Selective Patterns of Neurodegeneration In Vivo. *Cell Reports*, **2020**, 31, 107616	10.6	18
123	Utility of FDG-PET in diagnosis of Alzheimer-related TDP-43 proteinopathy. *Neurology*, **2020**, 95, e23-e34	6.5	11
122	Premature termination codon readthrough upregulates progranulin expression and improves lysosomal function in preclinical models of GRN deficiency. *Molecular Neurodegeneration*, **2020**, 15, 21	19	8
121	A Puts the Alpha in Synuclein. *Neuron*, **2020**, 105, 205-206	13.9	
120	Elevated methylation levels, reduced expression levels, and frequent contractions in a clinical cohort of C9orf72 expansion carriers. *Molecular Neurodegeneration*, **2020**, 15, 7	19	20
119	Posttranslational Modifications Mediate the Structural Diversity of Tauopathy Strains. *Cell*, **2020**, 180, 633-644.e12	56.2	156
118	Reduced C9ORF72 function exacerbates gain of toxicity from ALS/FTD-causing repeat expansion in C9orf72. *Nature Neuroscience*, **2020**, 23, 615-624	25.5	80
117	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. *Journal of Clinical Investigation*, **2020**, 130, 6080-6092	15.9	34
116	Chimeric Peptide Species Contribute to Divergent Dipeptide Repeat Pathology in c9ALS/FTD and SCA36. *Neuron*, **2020**, 107, 292-305.e6	13.9	25

115	Toward allele-specific targeting therapy and pharmacodynamic marker for spinocerebellar ataxia type 3. *Science Translational Medicine*, **2020**, 12,	17.5	15
114	Astrocyte-derived clusterin suppresses amyloid formation in vivo. *Molecular Neurodegeneration*, **2020**, 15, 71	19	11
113	Plasma neurofilament light predicts mortality in patients with stroke. *Science Translational Medicine* , **2020**, 12,	17.5	20
112	Clusterin ameliorates tau pathology in vivo by inhibiting fibril formation. *Acta Neuropathologica Communications*, **2020**, 8, 210	7.3	5
111	Structural Features of Small Molecules Targeting the RNA Repeat Expansion That Causes Genetically Defined ALS/FTD. *ACS Chemical Biology*, **2020**, 15, 3112-3123	4.9	4
110	poly(GR) aggregation induces TDP-43 proteinopathy. *Science Translational Medicine*, **2020**, 12,	17.5	49
109	Mitophagy alterations in Alzheimer's disease are associated with granulovacuolar degeneration and early tau pathology. *Alzheimers and Dementia*, **2020**, 17, 417	1.2	13
108	The influence of tau, amyloid, alpha-synuclein, TDP-43, and vascular pathology in clinically normal elderly individuals. *Neurobiology of Aging*, **2019**, 77, 26-36	5.6	32
107	Enhanced phosphorylation of T153 in soluble tau is a defining biochemical feature of the A152T tau risk variant. *Acta Neuropathologica Communications*, **2019**, 7, 10	7.3	1
106	Toxic expanded GGGGCC repeat transcription is mediated by the PAF1 complex in C9orf72-associated FTD. *Nature Neuroscience*, **2019**, 22, 863-874	25.5	38
105	Systematic analysis of dark and camouflaged genes reveals disease-relevant genes hiding in plain sight. *Genome Biology*, **2019**, 20, 97	18.3	68
104	eIF4B and eIF4H mediate GR production from expanded G4C2 in a Drosophila model for C9orf72-associated ALS. *Acta Neuropathologica Communications*, **2019**, 7, 62	7.3	22
103	Genetic Convergence Brings Clarity to the Enigmatic Red Line in ALS. *Neuron*, **2019**, 101, 1057-1069	13.9	63
102	Tau exhibits unique seeding properties in globular glial tauopathy. *Acta Neuropathologica Communications*, **2019**, 7, 36	7.3	14
101	ADAR2 mislocalization and widespread RNA editing aberrations in C9orf72-mediated ALS/FTD. *Acta Neuropathologica*, **2019**, 138, 49-65	14.3	27
100	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	14.3	50
99	Aberrant deposition of stress granule-resident proteins linked to C9orf72-associated TDP-43 proteinopathy. *Molecular Neurodegeneration*, **2019**, 14, 9	19	64
98	RPS25 is required for efficient RAN translation of C9orf72 and other neurodegenerative disease-associated nucleotide repeats. *Nature Neuroscience*, **2019**, 22, 1383-1388	25.5	54

97	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	7.3	9
96	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. *Acta Neuropathologica Communications*, **2019**, 7, 150	7.3	18
95	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1782-1796	5.3	11
94	Heterochromatin anomalies and double-stranded RNA accumulation underlie poly(PR) toxicity. *Science*, **2019**, 363,	33.3	104
93	Pathological, imaging and genetic characteristics support the existence of distinct TDP-43 types in non-FTLD brains. *Acta Neuropathologica*, **2019**, 137, 227-238	14.3	32
92	The Hairpin Form of r(GC) in c9ALS/FTD Is Repeat-Associated Non-ATG Translated and a Target for Bioactive Small Molecules. *Cell Chemical Biology*, **2019**, 26, 179-190.e12	8.2	43
91	TIA1 regulates the generation and response to toxic tau oligomers. *Acta Neuropathologica*, **2019**, 137, 259-277	14.3	39
90	Poly(GP), neurofilament and grey matter deficits in expansion carriers. *Annals of Clinical and Translational Neurology*, **2018**, 5, 583-597	5.3	29
89	Unaffected mosaic case: RNA foci, dipeptide proteins, but upregulated C9orf72 expression. *Neurology*, **2018**, 90, e323-e331	6.5	24
88	CUG initiation and frameshifting enable production of dipeptide repeat proteins from ALS/FTD C9ORF72 transcripts. *Nature Communications*, **2018**, 9, 152	17.4	77
87	TDP-43 pathology disrupts nuclear pore complexes and nucleocytoplasmic transport in ALS/FTD. *Nature Neuroscience*, **2018**, 21, 228-239	25.5	240
86	A zebrafish model for C9orf72 ALS reveals RNA toxicity as a pathogenic mechanism. *Acta Neuropathologica*, **2018**, 135, 427-443	14.3	66
85	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	24.1	60
84	Disease Mechanisms of Repeat Expansions. *Cold Spring Harbor Perspectives in Medicine*, **2018**, 8,	5.4	49
83	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	7.3	14
82	Replication of progressive supranuclear palsy genome-wide association study identifies SLCO1A2 and DUSP10 as new susceptibility loci. *Molecular Neurodegeneration*, **2018**, 13, 37	19	28
81	Microglial translational profiling reveals a convergent APOE pathway from aging, amyloid, and tau. *Journal of Experimental Medicine*, **2018**, 215, 2235-2245	16.6	85
80	Poly-GR dipeptide repeat polymers correlate with neurodegeneration and Clinicopathological subtypes in C9ORF72-related brain disease. *Acta Neuropathologica Communications*, **2018**, 6, 63	7.3	51

79	Dipeptide repeat proteins activate a heat shock response found in C9ORF72-ALS/FTLD patients. *Acta Neuropathologica Communications*, **2018**, 6, 55	7.3	15
78	Long-read sequencing across the C9orf72 'GGGGCC' repeat expansion: implications for clinical use and genetic discovery efforts in human disease. *Molecular Neurodegeneration*, **2018**, 13, 46	19	66
77	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,	4.8	8
76	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	3.5	10
75	Converging pathways in neurodegeneration, from genetics to mechanisms. *Nature Neuroscience*, **2018**, 21, 1300-1309	25.5	183
74	Association of Apolipoprotein E ε4 With Transactive Response DNA-Binding Protein 43. *JAMA Neurology*, **2018**, 75, 1347-1354	17.2	42
73	APOE ε4 is associated with increased tau pathology in primary tauopathy. *Nature Communications*, **2018**, 9, 4388	17.4	68
72	TRIO gene segregation in a family with cerebellar ataxia. *Neurologia I Neurochirurgia Polska*, **2018**, 52, 743-749	1	5
71	Tau Protein Disrupts Nucleocytoplasmic Transport in Alzheimer's Disease. *Neuron*, **2018**, 99, 925-940.e7	13.9	169
70	Poly(GR) impairs protein translation and stress granule dynamics in C9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis. *Nature Medicine*, **2018**, 24, 1136-1142	50.5	149
69	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	14.3	91
68	Spinal poly-GA inclusions in a C9orf72 mouse model trigger motor deficits and inflammation without neuron loss. *Acta Neuropathologica*, **2017**, 134, 241-254	14.3	70
67	Mutant TDP-43 does not impair mitochondrial bioenergetics in vitro and in vivo. *Molecular Neurodegeneration*, **2017**, 12, 37	19	28
66	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. *Acta Neuropathologica*, **2017**, 134, 255-269	14.3	57
65	Phosphorylated neurofilament heavy chain: A biomarker of survival for C9ORF72-associated amyotrophic lateral sclerosis. *Annals of Neurology*, **2017**, 82, 139-146	9.4	58
64	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	5.6	41
63	Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. *Science Translational Medicine*, **2017**, 9,	17.5	128
62	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	5.6	43

61	ARHGEF28 p.Lys280Metfs40Ter in an amyotrophic lateral sclerosis family with a C9orf72 expansion. *Neurology: Genetics*, **2017**, 3, e190	3.8	4
60	The lysosomal protein cathepsin L is a progranulin protease. *Molecular Neurodegeneration*, **2017**, 12, 55	19	54
59	An acetylation-phosphorylation switch that regulates tau aggregation propensity and function. *Journal of Biological Chemistry*, **2017**, 292, 15277-15286	5.4	78
58	TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. *Neuron*, **2017**, 95, 808-816.e9	13.9	341
57	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. *Human Molecular Genetics*, **2017**, 26, 3421-3431	5.6	63
56	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	11.5	66
55	Abnormal expression of homeobox genes and transthyretin in expansion carriers. *Neurology: Genetics*, **2017**, 3, e161	3.8	9
54	FTDP-17 with Pick body-like inclusions associated with a novel tau mutation, p.E372G. *Brain Pathology*, **2017**, 27, 612-626	6	11
53	An autoradiographic evaluation of AV-1451 Tau PET in dementia. *Acta Neuropathologica Communications*, **2016**, 4, 58	7.3	305
52	Timing and significance of pathological features in C9orf72 expansion-associated frontotemporal dementia. *Brain*, **2016**, 139, 3202-3216	11.2	90
51	Monitoring peripheral nerve degeneration in ALS by label-free stimulated Raman scattering imaging. *Nature Communications*, **2016**, 7, 13283	17.4	56
50	ALS and FTD: an epigenetic perspective. *Acta Neuropathologica*, **2016**, 132, 487-502	14.3	51
49	Interaction of tau with the RNA-Binding Protein TIA1 Regulates tau Pathophysiology and Toxicity. *Cell Reports*, **2016**, 15, 1455-1466	10.6	176
48	Updated TDP-43 in Alzheimer's disease staging scheme. *Acta Neuropathologica*, **2016**, 131, 571-85	14.3	168
47	TDP-43 functions within a network of hnRNP proteins to inhibit the production of a truncated human SORT1 receptor. *Human Molecular Genetics*, **2016**, 25, 534-45	5.6	52
46	C9ORF72 poly(GA) aggregates sequester and impair HR23 and nucleocytoplasmic transport proteins. *Nature Neuroscience*, **2016**, 19, 668-677	25.5	201
45	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. *Experimental Neurology*, **2016**, 277, 171-177	5.7	16
44	Gain of Toxicity from ALS/FTD-Linked Repeat Expansions in C9ORF72 Is Alleviated by Antisense Oligonucleotides Targeting GGGGCC-Containing RNAs. *Neuron*, **2016**, 90, 535-50	13.9	331

43	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	13.9	220
42	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. *Science*, **2016**, 353, 708-12	33.3	92
41	Insights into the pathogenic mechanisms of Chromosome 9 open reading frame 72 (C9orf72) repeat expansions. *Journal of Neurochemistry*, **2016**, 138 Suppl 1, 145-62	6	54
40	Understanding biomarkers of neurodegeneration: Novel approaches to detecting tau pathology. *Nature Medicine*, **2015**, 21, 219-20	50.5	14
39	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. *Nature Neuroscience*, **2015**, 18, 1175-82	25.5	235
38	Neurodegeneration. C9ORF72 repeat expansions in mice cause TDP-43 pathology, neuronal loss, and behavioral deficits. *Science*, **2015**, 348, 1151-4	33.3	279
37	Differential Toxicity of Nuclear RNA Foci versus Dipeptide Repeat Proteins in a Drosophila Model of C9ORF72 FTD/ALS. *Neuron*, **2015**, 87, 1207-1214	13.9	149
36	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. *Acta Neuropathologica*, **2015**, 130, 863-76	14.3	81
35	GGGGCC repeat expansion in C9orf72 compromises nucleocytoplasmic transport. *Nature*, **2015**, 525, 129-33	50.4	540
34	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. *Acta Neuropathologica*, **2015**, 130, 559-73	14.3	72
33	Quantitative analysis and clinico-pathological correlations of different dipeptide repeat protein pathologies in C9ORF72 mutation carriers. *Acta Neuropathologica*, **2015**, 130, 845-61	14.3	155
32	C9orf72 BAC Transgenic Mice Display Typical Pathologic Features of ALS/FTD. *Neuron*, **2015**, 88, 892-901	13.9	201
31	Human C9ORF72 Hexanucleotide Expansion Reproduces RNA Foci and Dipeptide Repeat Proteins but Not Neurodegeneration in BAC Transgenic Mice. *Neuron*, **2015**, 88, 902-909	13.9	183
30	Linking the VPS35 and EIF4G1 pathways in Parkinson's disease. *Neuron*, **2015**, 85, 1-3	13.9	29
29	Mechanisms of toxicity in C9FTLD/ALS. *Acta Neuropathologica*, **2014**, 127, 359-76	14.3	114
28	Characterization of DNA hypermethylation in the cerebellum of c9FTD/ALS patients. *Brain Research* , **2014**, 1584, 15-21	3.7	63
27	Severe amygdala dysfunction in a MAPT transgenic mouse model of frontotemporal dementia. *Neurobiology of Aging*, **2014**, 35, 1769-77	5.6	37
26	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. *Neurobiology of Aging*, **2014**, 35, 2421.e13-7	5.6	62

25	Discovery of a biomarker and lead small molecules to target r(GGGGCC)-associated defects in c9FTD/ALS. *Neuron*, **2014**, 83, 1043-50	13.9	232
24	Epigenetic modifications of theC9ORF72gene: a potential biomarker of disease?. *Future Neurology*, **2014**, 9, 123-126	1.5	
23	ER-mitochondria associations are regulated by the VAPB-PTPIP51 interaction and are disrupted by ALS/FTD-associated TDP-43. *Nature Communications*, **2014**, 5, 3996	17.4	341
22	Alterations in microRNA-124 and AMPA receptors contribute to social behavioral deficits in frontotemporal dementia. *Nature Medicine*, **2014**, 20, 1444-51	50.5	125
21	TDP-1, the Caenorhabditis elegans ortholog of TDP-43, limits the accumulation of double-stranded RNA. *EMBO Journal*, **2014**, 33, 2947-66	13	44
20	Aggregation-prone c9FTD/ALS poly(GA) RAN-translated proteins cause neurotoxicity by inducing ER stress. *Acta Neuropathologica*, **2014**, 128, 505-24	14.3	227
19	Expanded C9ORF72 hexanucleotide repeat in depressive pseudodementia. *JAMA Neurology*, **2014**, 71, 775-81	17.2	24
18	Divergent phenotypes in mutant TDP-43 transgenic mice highlight potential confounds in TDP-43 transgenic modeling. *PLoS ONE*, **2014**, 9, e86513	3.7	23
17	Homozygosity for the C9orf72 GGGGCC repeat expansion in frontotemporal dementia. *Acta Neuropathologica*, **2013**, 126, 401-9	14.3	119
16	Targeting RNA foci in iPSC-derived motor neurons from ALS patients with a C9ORF72 repeat expansion. *Science Translational Medicine*, **2013**, 5, 208ra149	17.5	488
15	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, 978-88	24.1	200
14	Unconventional translation of C9ORF72 GGGGCC expansion generates insoluble polypeptides specific to c9FTD/ALS. *Neuron*, **2013**, 77, 639-46	13.9	783
13	RNA toxicity from the ALS/FTD C9ORF72 expansion is mitigated by antisense intervention. *Neuron*, **2013**, 80, 415-28	13.9	650
12	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, 829-44	14.3	392
11	Reduced C9orf72 gene expression in c9FTD/ALS is caused by histone trimethylation, an epigenetic event detectable in blood. *Acta Neuropathologica*, **2013**, 126, 895-905	14.3	217
10	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. *Neurology*, **2013**, 81, 1332-41	6.5	75
9	The dual functions of the extreme N-terminus of TDP-43 in regulating its biological activity and inclusion formation. *Human Molecular Genetics*, **2013**, 22, 3112-22	5.6	119
8	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, 21510-5	11.5	61

7	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, 10851-9	6.6	373
6	O1-07-01: Accelerated lipofuscinosis and ubiquitination in granulin knockout mice suggests a role for progranulin in successful aging **2010**, 6, S83-S83		
5	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, 7607-12	11.5	433
4	Novel mutations in TARDBP (TDP-43) in patients with familial amyotrophic lateral sclerosis. *PLoS Genetics*, **2008**, 4, e1000193	6	339
3	Identification and characterization of the human parkin gene promoter. *Journal of Neurochemistry*, **2001**, 78, 1146-52	6	25
2	Lewy bodies and parkinsonism in families with parkin mutations. *Annals of Neurology*, **2001**, 50, 293-300	9.4	425
1	Long-read sequencing across the C9orf72 □GGGGCC□repeat expansion: implications for clinical use and genetic discovery efforts in human disease		1