Rosa Rademakers

List of Publications by Year
in descending order

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279	39,326	4658 85	3261 185
papers	citations	h-index	g-index

298	298	298	25532
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Expanded GGGGCC Hexanucleotide Repeat in Noncoding Region of C9ORF72 Causes Chromosome 9p-Linked FTD and ALS. Neuron, 2011, 72, 245-256.	8.1	4,176
2	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	27.0	2,385
3	Mutations in progranulin cause tau-negative frontotemporal dementia linked to chromosome 17. Nature, 2006, 442, 916-919.	27.8	1,816
4	Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21. Nature, 2006, 442, 920-924.	27.8	1,386
5	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	27.8	1,249
6	Unconventional Translation of C9ORF72 GGGGCC Expansion Generates Insoluble Polypeptides Specific to c9FTD/ALS. Neuron, 2013, 77, 639-646.	8.1	962
7	Limbic-predominant age-related TDP-43 encephalopathy (LATE): consensus working group report. Brain, 2019, 142, 1503-1527.	7.6	873
8	TDP-43 and FUS in amyotrophic lateral sclerosis and frontotemporal dementia. Lancet Neurology, The, 2010, 9, 995-1007.	10.2	816
9	A new subtype of frontotemporal lobar degeneration with FUS pathology. Brain, 2009, 132, 2922-2931.	7.6	628
10	Progranulin Deficiency Promotes Circuit-Specific Synaptic Pruning by Microglia via Complement Activation. Cell, 2016, 165, 921-935.	28.9	558
11	Mutations in progranulin are a major cause of ubiquitin-positive frontotemporal lobar degeneration. Human Molecular Genetics, 2006, 15, 2988-3001.	2.9	529
12	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
13	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.	7.7	506
14	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705.	21.4	502
15	TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. Neuron, 2017, 95, 808-816.e9.	8.1	493
16	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	21.4	479
17	Mutations in the colony stimulating factor 1 receptor (CSF1R) gene cause hereditary diffuse leukoencephalopathy with spheroids. Nature Genetics, 2012, 44, 200-205.	21.4	428
18	TDP-43 pathology disrupts nuclear pore complexes and nucleocytoplasmic transport in ALS/FTD. Nature Neuroscience, 2018, 21, 228-239.	14.8	404

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19	Apolipoprotein E Is a Ligand for Triggering Receptor Expressed on Myeloid Cells 2 (TREM2). Journal of Biological Chemistry, 2015, 290, 26043-26050.	3.4	395
20	Novel Mutations in TARDBP (TDP-43) in Patients with Familial Amyotrophic Lateral Sclerosis. PLoS Genetics, 2008, 4, e1000193.	3.5	393
21	Neuroimaging signatures of frontotemporal dementia genetics: C9ORF72, tau, progranulin and sporadics. Brain, 2012, 135, 794-806.	7.6	355
22	Advances in understanding the molecular basis of frontotemporal dementia. Nature Reviews Neurology, 2012, 8, 423-434.	10.1	353
23	Plasma progranulin levels predict progranulin mutation status in frontotemporal dementia patients and asymptomatic family members. Brain, 2009, 132, 583-591.	7.6	344
24	C9ORF72 repeat expansions in mice cause TDP-43 pathology, neuronal loss, and behavioral deficits. Science, 2015, 348, 1151-1154.	12.6	332
25	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. Nature Neuroscience, 2015, 18, 1175-1182.	14.8	330
26	TREM2 in neurodegeneration: evidence for association of the p.R47H variant with frontotemporal dementia and Parkinsonâ€™s disease. Molecular Neurodegeneration, 2013, 8, 19.	10.8	323
27	Characterization of frontotemporal dementia and/or amyotrophic lateral sclerosis associated with the GGGGCC repeat expansion in C9ORF72. Brain, 2012, 135, 765-783.	7.6	322
28	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	10.2	302
29	The neuropathology of frontotemporal lobar degeneration caused by mutations in the progranulin gene. Brain, 2006, 129, 3081-3090.	7.6	291
30	Discovery of a Biomarker and Lead Small Molecules to Target r(GGGGCC)-Associated Defects in c9FTD/ALS. Neuron, 2014, 83, 1043-1050.	8.1	289
31	Aggregation-prone c9FTD/ALS poly(GA) RAN-translated proteins cause neurotoxicity by inducing ER stress. Acta Neuropathologica, 2014, 128, 505-524.	7.7	284
32	Clinical and neuropathologic heterogeneity of c9FTD/ALS associated with hexanucleotide repeat expansion in C9ORF72. Acta Neuropathologica, 2011, 122, 673-690.	7.7	277
33	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	5.5	277
34	Common variation in the miR-659 binding-site of GRN is a major risk factor for TDP43-positive frontotemporal dementia. Human Molecular Genetics, 2008, 17, 3631-3642.	2.9	271
35	Corticobasal degeneration: a pathologically distinct 4R tauopathy. Nature Reviews Neurology, 2011, 7, 263-272.	10.1	270
36	C9ORF72 poly(GA) aggregates sequester and impair HR23 and nucleocytoplasmic transport proteins. Nature Neuroscience, 2016, 19, 668-677.	14.8	268

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37	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. Acta Neuropathologica, 2015, 130, 77-92.	7.7	267
38	Reduced C9orf72 gene expression in c9FTD/ALS is caused by histone trimethylation, an epigenetic event detectable in blood. Acta Neuropathologica, 2013, 126, 895-905.	7.7	263
39	FET proteins TAF15 and EWS are selective markers that distinguish FTLD with FUS pathology from amyotrophic lateral sclerosis with FUS mutations. Brain, 2011, 134, 2595-2609.	7.6	247
40	Poly(GR) impairs protein translation and stress granule dynamics in C9orf72-associated frontotemporal dementia and amyotrophic lateral sclerosis. Nature Medicine, 2018, 24, 1136-1142.	30.7	241
41	Chronic traumatic encephalopathy pathology in a neurodegenerative disorders brain bank. Acta Neuropathologica, 2015, 130, 877-889.	7.7	235
42	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.	10.2	232
43	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
44	FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. Acta Neuropathologica, 2010, 120, 33-41.	7.7	222
45	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
46	Phenotypic variability associated with progranulin haploinsufficiency in patients with the common 1477Câ†'T (Arg493X) mutation: an international initiative. Lancet Neurology, The, 2007, 6, 857-868.	10.2	199
47	Evidence for a role of the rare p.A152T variant in MAPT in increasing the risk for FTD-spectrum and Alzheimer's diseases. Human Molecular Genetics, 2012, 21, 3500-3512.	2.9	198
48	Neuropathologic Features of Frontotemporal Lobar Degeneration With Ubiquitin-Positive Inclusions With Progranulin Gene (PGRN) Mutations. Journal of Neuropathology and Experimental Neurology, 2007, 66, 142-151.	1.7	184
49	Frontotemporal dementia due to C9ORF72 mutations. Neurology, 2012, 79, 1002-1011.	1.1	183
50	Heterochromatin anomalies and double-stranded RNA accumulation underlie C9orf72 poly(PR) toxicity. Science, 2019, 363, .	12.6	181
51	Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72 -associated amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	179
52	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. Lancet Neurology, The, 2020, 19, 145-156.	10.2	175
53	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
54	Clinical, neuroimaging and neuropathological features of a new chromosome 9p-linked FTD-ALS family. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 196-203.	1.9	170

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55	Genome-wide association study of corticobasal degeneration identifies risk variants shared with progressive supranuclear palsy. Nature Communications, 2015, 6, 7247.	12.8	170
56	How do C9ORF72 repeat expansions cause amyotrophic lateral sclerosis and frontotemporal dementia. Current Opinion in Neurology, 2012, 25, 689-700.	3.6	169
57	High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. Human Molecular Genetics, 2005, 14, 3281-3292.	2.9	156
58	MRI characteristics and scoring in HDLS due to CSF1R gene mutations. Neurology, 2012, 79, 566-574.	1.1	153
59	Patterns of gray matter atrophy in genetic frontotemporal dementia: results from the GENFI study. Neurobiology of Aging, 2018, 62, 191-196.	3.1	151
60	The role of transactive response DNA-binding protein-43 in amyotrophic lateral sclerosis and frontotemporal dementia. Current Opinion in Neurology, 2008, 21, 693-700.	3.6	150
61	Ataxin-2 repeat-length variation and neurodegeneration. Human Molecular Genetics, 2011, 20, 3207-3212.	2.9	147
62	De novo truncating FUS gene mutation as a cause of sporadic amyotrophic lateral sclerosis. Human Mutation, 2010, 31, E1377-E1389.	2.5	141
63	Altered network connectivity in frontotemporal dementia with C9orf72 hexanucleotide repeat expansion. Brain, 2014, 137, 3047-3060.	7.6	140
64	TARDBP 3â€²-UTR variant in autopsy-confirmed frontotemporal lobar degeneration with TDP-43 proteinopathy. Acta Neuropathologica, 2009, 118, 633-645.	7.7	139
65	TDP-43 frontotemporal lobar degeneration and autoimmune disease. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 956-962.	1.9	137
66	CSF1R mutations link POLD and HDLS as a single disease entity. Neurology, 2013, 80, 1033-1040.	1.1	136
67	TMEM106B protects C9ORF72 expansion carriers against frontotemporal dementia. Acta Neuropathologica, 2014, 127, 397-406.	7.7	133
68	Genome-wide Screen Identifies rs646776 near Sortilin as a Regulator of Progranulin Levels in Human Plasma. American Journal of Human Genetics, 2010, 87, 890-897.	6.2	130
69	Progranulin regulates neuronal outgrowth independent of Sortilin. Molecular Neurodegeneration, 2012, 7, 33.	10.8	129
70	Network degeneration and dysfunction in presymptomatic C9ORF72 expansion carriers. NeuroImage: Clinical, 2017, 14, 286-297.	2.7	129
71	Systematic analysis of dark and camouflaged genes reveals disease-relevant genes hiding in plain sight. Genome Biology, 2019, 20, 97.	8.8	122
72	Neuropathological features of corticobasal degeneration presenting as corticobasal syndrome or Richardson syndrome. Brain, 2011, 134, 3264-3275.	7.6	119

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73	Parkinsonian features in hereditary diffuse leukoencephalopathy with spheroids (HDLS) and CSF1R mutations. Parkinsonism and Related Disorders, 2013, 19, 869-877.	2.2	119
74	Differential clinicopathologic and genetic features of late-onset amnestic dementias. Acta Neuropathologica, 2014, 128, 411-421.	7.7	119
75	Validation of serum neurofilaments as prognostic and potential pharmacodynamic biomarkers for ALS. Neurology, 2020, 95, e59-e69.	1.1	119
76	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions inC9orf72reveals marked differences in results among 14 laboratories. Journal of Medical Genetics, 2014, 51, 419-424.	3.2	118
77	Genetics of <scp>FTLD</scp>: overview and what else we can expect from genetic studies. Journal of Neurochemistry, 2016, 138, 32-53.	3.9	118
78	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	8.2	117
79	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. Science, 2016, 353, 708-712.	12.6	116
80	The chromosome 9 ALS and FTD locus is probably derived from a single founder. Neurobiology of Aging, 2012, 33, 209.e3-209.e8.	3.1	115
81	TREM2 in CNS homeostasis and neurodegenerative disease. Molecular Neurodegeneration, 2015, 10, 43.	10.8	115
82	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.	10.8	111
83	Genetic and Clinical Features of Progranulin-Associated Frontotemporal Lobar Degeneration. Archives of Neurology, 2011, 68, 488.	4.5	108
84	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. Acta Neuropathologica, 2015, 130, 863-876.	7.7	104
85	Fus gene mutations in familial and sporadic amyotrophic lateral sclerosis. Muscle and Nerve, 2010, 42, 170-176.	2.2	101
86	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. Human Molecular Genetics, 2017, 26, 3421-3431.	2.9	101
87	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	14.8	101
88	Regional distribution of synaptic markers and APP correlate with distinct clinicopathological features in sporadic and familial Alzheimerâ€™s disease. Brain, 2014, 137, 1533-1549.	7.6	100
89	APOE Îµ2 is associated with increased tau pathology in primary tauopathy. Nature Communications, 2018, 9, 4388.	12.8	100
90	Lipidomic and Transcriptomic Basis of Lysosomal Dysfunction in Progranulin Deficiency. Cell Reports, 2017, 20, 2565-2574.	6.4	98

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91	Frontotemporal dementia: a bridge between dementia and neuromuscular disease. Annals of the New York Academy of Sciences, 2015, 1338, 71-93.	3.8	97
92	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.	10.2	97
93	Shared genetic risk between corticobasal degeneration, progressive supranuclear palsy, and frontotemporal dementia. Acta Neuropathologica, 2017, 133, 825-837.	7.7	90
94	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.	7.7	90
95	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. Acta Neuropathologica, 2015, 130, 559-573.	7.7	89
96	A Belgian ancestral haplotype harbours a highly prevalent mutation for 17q21-linked tau-negative FTLD. Brain, 2006, 129, 841-852.	7.6	88
97	Tau pathology in frontotemporal lobar degeneration with C9ORF72 hexanucleotide repeat expansion. Acta Neuropathologica, 2013, 125, 289-302.	7.7	87
98	<scp>TMEM</scp>106B p.T185S regulates <scp>TMEM</scp>106B protein levels: implications for frontotemporal dementia. Journal of Neurochemistry, 2013, 126, 781-791.	3.9	87
99	A nonsynonymous mutation in PLCG2 reduces the risk of Alzheimerâ€™s disease, dementia with Lewy bodies and frontotemporal dementia, and increases the likelihood of longevity. Acta Neuropathologica, 2019, 138, 237-250.	7.7	87
100	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
101	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. Neurology, 2013, 81, 1332-1341.	1.1	84
102	Length of normal alleles of C9ORF72 GGGGCC repeat do not influence disease phenotype. Neurobiology of Aging, 2012, 33, 2950.e5-2950.e7.	3.1	83
103	What we know about TMEM106B in neurodegeneration. Acta Neuropathologica, 2016, 132, 639-651.	7.7	83
104	Genomic architecture of human 17q21 linked to frontotemporal dementia uncovers a highly homologous family of low-copy repeats in the tau region. Human Molecular Genetics, 2005, 14, 1753-1762.	2.9	82
105	Common Variant in <i>GRN</i> Is a Genetic Risk Factor for Hippocampal Sclerosis in the Elderly. Neurodegenerative Diseases, 2010, 7, 170-174.	1.4	82
106	TMEM106B risk variant is implicated in the pathologic presentation of Alzheimer disease. Neurology, 2012, 79, 717-718.	1.1	81
107	Poly-GR dipeptide repeat polymers correlate with neurodegeneration and Clinicopathological subtypes in C9ORF72-related brain disease. Acta Neuropathologica Communications, 2018, 6, 63.	5.2	79
108	Hippocampal Sclerosis in the Elderly. Alzheimer Disease and Associated Disorders, 2011, 25, 364-368.	1.3	78

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109	Increased prevalence of autoimmune disease within C9 and FTD/MND cohorts. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e301.	6.0	78
110	rs5848 polymorphism and serum progranulin level. Journal of the Neurological Sciences, 2011, 300, 28-32.	0.6	77
111	The molecular genetics and neuropathology of frontotemporal lobar degeneration: recent developments. Neurogenetics, 2007, 8, 237-248.	1.4	76
112	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. Acta Neuropathologica, 2017, 134, 255-269.	7.7	76
113	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. Neurobiology of Aging, 2014, 35, 2421.e13-2421.e17.	3.1	74
114	Progranulin protein levels are differently regulated in plasma and CSF. Neurology, 2014, 82, 1871-1878.	1.1	70
115	Homotypic fibrillization of TMEM106B across diverse neurodegenerative diseases. Cell, 2022, 185, 1346-1355.e15.	28.9	70
116	Progranulin-associated PiB-negative logopenic primary progressive aphasia. Journal of Neurology, 2014, 261, 604-614.	3.6	69
117	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	3.1	69
118	Prosaposin is a regulator of progranulin levels and oligomerization. Nature Communications, 2016, 7, 11992.	12.8	68
119	Hippocampal sclerosis in Lewy body disease is a TDP-43 proteinopathy similar to FTLD-TDP Type A. Acta Neuropathologica, 2015, 129, 53-64.	7.7	67
120	Novel mutation in MAPT exon 13 (p.N410H) causes corticobasal degeneration. Acta Neuropathologica, 2014, 127, 271-282.	7.7	66
121	Pathological, imaging and genetic characteristics support the existence of distinct TDP-43 types in non-FTLD brains. Acta Neuropathologica, 2019, 137, 227-238.	7.7	65
122	A Point Mutation in PDGFRB Causes Autosomal-Dominant Penttinen Syndrome. American Journal of Human Genetics, 2015, 97, 465-474.	6.2	64
123	Genetic modifiers in carriers of repeat expansions in the C9ORF72 gene. Molecular Neurodegeneration, 2014, 9, 38.	10.8	63
124	Expression of Fused in sarcoma mutations in mice recapitulates the neuropathology of FUS proteinopathies and provides insight into disease pathogenesis. Molecular Neurodegeneration, 2012, 7, 53.	10.8	61
125	Cerebellar ataxia in progressive supranuclear palsy: An autopsy study of PSPâ€€. Movement Disorders, 2016, 31, 653-662.	3.9	60
126	In vivo ¹⁸ F-AV-1451 tau PET signal in MAPT mutation carriers varies by expected tau isoforms. Neurology, 2018, 90, e947-e954.	1.1	60

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127	Alzheimer Diseaseâ€“like Phenotype Associated With the c.154delA Mutation in Progranulin. Archives of Neurology, 2010, 67, 171-7.	4.5	59
128	Sporadic corticobasal syndrome due to FTLD-TDP. Acta Neuropathologica, 2010, 119, 365-374.	7.7	59
129	Frontotemporal dementia-associated N279K tau mutant disrupts subcellular vesicle trafficking and induces cellular stress in iPSC-derived neural stem cells. Molecular Neurodegeneration, 2015, 10, 46.	10.8	58
130	SLC20A2 and THAP1 deletion in familial basal ganglia calcification with dystonia. Neurogenetics, 2014, 15, 23-30.	1.4	56
131	The neuropsychology of normal aging and preclinical Alzheimer's disease. Alzheimer's and Dementia, 2014, 10, 84-92.	0.8	55
132	Cognitive reserve and TMEM106B genotype modulate brain damage in presymptomatic frontotemporal dementia: a GENFI study. Brain, 2017, 140, 1784-1791.	7.6	55
133	Replication of progressive supranuclear palsy genome-wide association study identifies SLCO1A2 and DUSP10 as new susceptibility loci. Molecular Neurodegeneration, 2018, 13, 37.	10.8	54
134	Plasma Neurofilament Light for Prediction of Disease Progression in Familial Frontotemporal Lobar Degeneration. Neurology, 2021, 96, e2296-e2312.	1.1	52
135	Recent insights into the molecular genetics of dementia. Trends in Neurosciences, 2009, 32, 451-461.	8.6	51
136	Jump from Pre-mutation to Pathologic Expansion in C9orf72. American Journal of Human Genetics, 2015, 96, 962-970.	6.2	50
137	Ossified blood vessels in primary familial brain calcification elicit a neurotoxic astrocyte response. Brain, 2019, 142, 885-902.	7.6	50
138	Clinical and neuroimaging biomarkers of amyloid-negative logopenic primary progressive aphasia. Brain and Language, 2015, 142, 45-53.	1.6	49
139	Linkage and Association Studies Identify a Novel Locus for Alzheimer Disease at 7q36 in a Dutch Population-Based Sample. American Journal of Human Genetics, 2005, 77, 643-652.	6.2	48
140	Altered microRNA expression in frontotemporal lobar degeneration with TDP-43 pathology caused by progranulin mutations. BMC Genomics, 2011, 12, 527.	2.8	48
141	<scp>S</scp>tudy of LRRK2 variation in tauopathy: Progressive supranuclear palsy and corticobasal degeneration. Movement Disorders, 2017, 32, 115-123.	3.9	48
142	Poly(GP), neurofilament and grey matter deficits in C9orf72 expansion carriers. Annals of Clinical and Translational Neurology, 2018, 5, 583-597.	3.7	48
143	C9orf72 repeat expansions in patients with ALS and FTD. Lancet Neurology, The, 2012, 11, 297-298.	10.2	46
144	Distribution and characteristics of transactive response DNA binding protein 43 kDa pathology in progressive supranuclear palsy. Movement Disorders, 2017, 32, 246-255.	3.9	46

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145	Cognitive impairment in progressive supranuclear palsy is associated with tau burden. Movement Disorders, 2017, 32, 1772-1779.	3.9	46
146	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
147	Frontotemporal lobar degeneration with TDPâ€43 proteinopathy and chromosome 9p repeat expansion in C9ORF72: clinicopathologic correlation. Neuropathology, 2013, 33, 122-133.	1.2	45
148	Genetic Screening and Functional Characterization ofPDGFRBMutations Associated with Basal Ganglia Calcification of Unknown Etiology. Human Mutation, 2014, 35, 964-971.	2.5	45
149	Loss of TMEM106B leads to myelination deficits: implications for frontotemporal dementia treatment strategies. Brain, 2020, 143, 1905-1919.	7.6	44
150	The genetics of frontotemporal lobar degeneration. Current Neurology and Neuroscience Reports, 2007, 7, 434-442.	4.2	43
151	TARDBP Mutation Analysis in TDP-43 Proteinopathies and Deciphering the Toxicity of Mutant TDP-43. Journal of Alzheimer's Disease, 2012, 33, S35-S45.	2.6	43
152	Three sib-pairs of autopsy-confirmed progressive supranuclear palsy. Parkinsonism and Related Disorders, 2015, 21, 101-105.	2.2	42
153	Loss of homeostatic microglial phenotype in CSF1R-related Leukoencephalopathy. Acta Neuropathologica Communications, 2020, 8, 72.	5.2	42
154	Genetics of Early-Onset Alzheimer Dementia. Scientific World Journal, The, 2003, 3, 497-519.	2.1	40
155	Analysis of COQ2gene in multiple system atrophy. Molecular Neurodegeneration, 2014, 9, 44.	10.8	40
156	Identical twins with the C9orf72 repeat expansion are discordant for ALS. Neurology, 2014, 83, 1476-1478.	1.1	40
157	Hereditary diffuse leukoencephalopathy with spheroids with phenotype of primary progressive multiple sclerosis. European Journal of Neurology, 2015, 22, 328-333.	3.3	40
158	Conserved DNA methylation combined with differential frontal cortex and cerebellar expression distinguishes C9orf72-associated and sporadic ALS, and implicates SERPINA1 in disease. Acta Neuropathologica, 2017, 134, 715-728.	7.7	40
159	Progranulin plasma levels predict the presence of GRN mutations in asymptomatic subjects and do not correlate with brain atrophy: results from the GENFI study. Neurobiology of Aging, 2018, 62, 245.e9-245.e12.	3.1	40
160	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. Acta Neuropathologica Communications, 2019, 7, 150.	5.2	40
161	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. Brain, 2018, 141, 2895-2907.	7.6	39
162	Presymptomatic white matter integrity loss in familial frontotemporal dementia in the <scp>GENFI</scp> cohort: A crossâ€sectional diffusion tensor imaging study. Annals of Clinical and Translational Neurology, 2018, 5, 1025-1036.	3.7	39

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163	Association of MAPT Subhaplotypes With Risk of Progressive Supranuclear Palsy and Severity of Tau Pathology. JAMA Neurology, 2019, 76, 710.	9.0	39
164	A novel tau mutation, p.K317N, causes globular glial tauopathy. Acta Neuropathologica, 2015, 130, 199-214.	7.7	38
165	Clinical and neuropathological features of ALS/FTD with TIA1 mutations. Acta Neuropathologica Communications, 2017, 5, 96.	5.2	38
166	Analysis of the C9orf72 repeat in Parkinson's disease, essential tremor and restless legs syndrome. Parkinsonism and Related Disorders, 2013, 19, 198-201.	2.2	37
167	Progranulin gene mutation with an unusual clinical and neuropathologic presentation. Movement Disorders, 2008, 23, 1168-1173.	3.9	36
168	Mitotic defects lead to neuronal aneuploidy and apoptosis in frontotemporal lobar degeneration caused by MAPT mutations. Molecular Biology of the Cell, 2018, 29, 575-586.	2.1	36
169	Genetic Disorders with Tau Pathology: A Review of the Literature and Report of Two Patients with Tauopathy and Positive Family Histories. Neurodegenerative Diseases, 2016, 16, 12-21.	1.4	35
170	Loss of Tmem106b exacerbates <scp>FTLD</scp> pathologies and causes motor deficits in progranulinâ€deficient mice. EMBO Reports, 2020, 21, e50197.	4.5	35
171	Network-driven plasma proteomics expose molecular changes in the Alzheimerâ€™s brain. Molecular Neurodegeneration, 2016, 11, 31.	10.8	34
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