Rosa Rademakers

List of Publications by Year in descending order

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#	Article	lF	CITATIONS
1	Expanded GGGGCC Hexanucleotide Repeat in Noncoding Region of C9ORF72 Causes Chromosome 9p-Linked FTD and ALS. Neuron, 2011, 72, 245-256.	3.8	4,176
2	<i>TREM2</i> Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	13.9	2,385
3	Mutations in progranulin cause tau-negative frontotemporal dementia linked to chromosome 17. Nature, 2006, 442, 916-919.	13.7	1,816
4	Null mutations in progranulin cause ubiquitin-positive frontotemporal dementia linked to chromosome 17q21. Nature, 2006, 442, 920-924.	13.7	1,386
5	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	13.7	1,249
6	Unconventional Translation of C9ORF72 GGGGCC Expansion Generates Insoluble Polypeptides Specific to c9FTD/ALS. Neuron, 2013, 77, 639-646.	3.8	962
7	Limbic-predominant age-related TDP-43 encephalopathy (LATE): consensus working group report. Brain, 2019, 142, 1503-1527.	3.7	873
8	TDP-43 and FUS in amyotrophic lateral sclerosis and frontotemporal dementia. Lancet Neurology, The, 2010, 9, 995-1007.	4.9	816
9	A new subtype of frontotemporal lobar degeneration with FUS pathology. Brain, 2009, 132, 2922-2931.	3.7	628
10	Progranulin Deficiency Promotes Circuit-Specific Synaptic Pruning by Microglia via Complement Activation. Cell, 2016, 165, 921-935.	13.5	558
11	Mutations in progranulin are a major cause of ubiquitin-positive frontotemporal lobar degeneration. Human Molecular Genetics, 2006, 15, 2988-3001.	1.4	529
12	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	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.	3.9	506
14	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705.	9.4	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.	3.8	493
16	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	9.4	479
17	Mutations in the colony stimulating factor 1 receptor (CSF1R) gene cause hereditary diffuse leukoencephalopathy with spheroids. Nature Genetics, 2012, 44, 200-205.	9.4	428
18	TDP-43 pathology disrupts nuclear pore complexes and nucleocytoplasmic transport in ALS/FTD. Nature Neuroscience, 2018, 21, 228-239.	7.1	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.	1.6	395
20	Novel Mutations in TARDBP (TDP-43) in Patients with Familial Amyotrophic Lateral Sclerosis. PLoS Genetics, 2008, 4, e1000193.	1.5	393
21	Neuroimaging signatures of frontotemporal dementia genetics: C9ORF72, tau, progranulin and sporadics. Brain, 2012, 135, 794-806.	3.7	355
22	Advances in understanding the molecular basis of frontotemporal dementia. Nature Reviews Neurology, 2012, 8, 423-434.	4.9	353
23	Plasma progranulin levels predict progranulin mutation status in frontotemporal dementia patients and asymptomatic family members. Brain, 2009, 132, 583-591.	3.7	344
24	<i>C9ORF72</i> repeat expansions in mice cause TDP-43 pathology, neuronal loss, and behavioral deficits. Science, 2015, 348, 1151-1154.	6.0	332
25	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. Nature Neuroscience, 2015, 18, 1175-1182.	7.1	330
26	TREM2 in neurodegeneration: evidence for association of the p.R47H variant with frontotemporal dementia and Parkinson's disease. Molecular Neurodegeneration, 2013, 8, 19.	4.4	323
27	Characterization of frontotemporal dementia and/or amyotrophic lateral sclerosis associated with the GGGGCC repeat expansion in C9ORF72. Brain, 2012, 135, 765-783.	3.7	322
28	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	4.9	302
29	The neuropathology of frontotemporal lobar degeneration caused by mutations in the progranulin gene. Brain, 2006, 129, 3081-3090.	3.7	291
30	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
31	Aggregation-prone c9FTD/ALS poly(GA) RAN-translated proteins cause neurotoxicity by inducing ER stress. Acta Neuropathologica, 2014, 128, 505-524.	3.9	284
32	Clinical and neuropathologic heterogeneity of c9FTD/ALS associated with hexanucleotide repeat expansion in C9ORF72. Acta Neuropathologica, 2011, 122, 673-690.	3.9	277
33	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	2.4	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.	1.4	271
35	Corticobasal degeneration: a pathologically distinct 4R tauopathy. Nature Reviews Neurology, 2011, 7, 263-272.	4.9	270
36	C9ORF72 poly(GA) aggregates sequester and impair HR23 and nucleocytoplasmic transport proteins. Nature Neuroscience, 2016, 19, 668-677.	7.1	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.	3.9	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.	3.9	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.	3.7	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.	15.2	241
41	Chronic traumatic encephalopathy pathology in a neurodegenerative disorders brain bank. Acta Neuropathologica, 2015, 130, 877-889.	3.9	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.	4.9	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.	9.4	223
44	FUS pathology defines the majority of tau- and TDP-43-negative frontotemporal lobar degeneration. Acta Neuropathologica, 2010, 120, 33-41.	3.9	222
45	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
46	Phenotypic variability associated with progranulin haploinsufficiency in patients with the common 1477C→T (Arg493X) mutation: an international initiative. Lancet Neurology, The, 2007, 6, 857-868.	4.9	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.	1.4	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.	0.9	184
49	Frontotemporal dementia due to <i>C9ORF72</i> mutations. Neurology, 2012, 79, 1002-1011.	1.5	183
50	Heterochromatin anomalies and double-stranded RNA accumulation underlie <i>C9orf72</i> poly(PR) toxicity. Science, 2019, 363, .	6.0	181
51	Poly(GP) proteins are a useful pharmacodynamic marker for <i>C9ORF72</i> -associated amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	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.	4.9	175
53	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	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.	0.9	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.	5.8	170
56	How do C9ORF72 repeat expansions cause amyotrophic lateral sclerosis and frontotemporal dementia. Current Opinion in Neurology, 2012, 25, 689-700.	1.8	169
57	High-density SNP haplotyping suggests altered regulation of tau gene expression in progressive supranuclear palsy. Human Molecular Genetics, 2005, 14, 3281-3292.	1.4	156
58	MRI characteristics and scoring in HDLS due to <i>CSF1R</i> gene mutations. Neurology, 2012, 79, 566-574.	1.5	153
59	Patterns of gray matter atrophy in genetic frontotemporal dementia: results from the GENFI study. Neurobiology of Aging, 2018, 62, 191-196.	1.5	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.	1.8	150
61	Ataxin-2 repeat-length variation and neurodegeneration. Human Molecular Genetics, 2011, 20, 3207-3212.	1.4	147
62	De novo truncating FUS gene mutation as a cause of sporadic amyotrophic lateral sclerosis. Human Mutation, 2010, 31, E1377-E1389.	1.1	141
63	Altered network connectivity in frontotemporal dementia with C9orf72 hexanucleotide repeat expansion. Brain, 2014, 137, 3047-3060.	3.7	140
64	TARDBP 3′-UTR variant in autopsy-confirmed frontotemporal lobar degeneration with TDP-43 proteinopathy. Acta Neuropathologica, 2009, 118, 633-645.	3.9	139
65	TDP-43 frontotemporal lobar degeneration and autoimmune disease. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 956-962.	0.9	137
66	<i>CSF1R</i> mutations link POLD and HDLS as a single disease entity. Neurology, 2013, 80, 1033-1040.	1.5	136
67	TMEM106B protects C9ORF72 expansion carriers against frontotemporal dementia. Acta Neuropathologica, 2014, 127, 397-406.	3.9	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.	2.6	130
69	Progranulin regulates neuronal outgrowth independent of Sortilin. Molecular Neurodegeneration, 2012, 7, 33.	4.4	129
70	Network degeneration and dysfunction in presymptomatic C9ORF72 expansion carriers. NeuroImage: Clinical, 2017, 14, 286-297.	1.4	129
71	Systematic analysis of dark and camouflaged genes reveals disease-relevant genes hiding in plain sight. Genome Biology, 2019, 20, 97.	3.8	122
72	Neuropathological features of corticobasal degeneration presenting as corticobasal syndrome or Richardson syndrome. Brain, 2011, 134, 3264-3275.	3.7	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.	1.1	119
74	Differential clinicopathologic and genetic features of late-onset amnestic dementias. Acta Neuropathologica, 2014, 128, 411-421.	3.9	119
75	Validation of serum neurofilaments as prognostic and potential pharmacodynamic biomarkers for ALS. Neurology, 2020, 95, e59-e69.	1.5	119
76	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in <i>C9orf72</i> reveals marked differences in results among 14 laboratories. Journal of Medical Genetics, 2014, 51, 419-424.	1.5	118
77	Genetics of <scp>FTLD</scp> : overview and what else we can expect from genetic studies. Journal of Neurochemistry, 2016, 138, 32-53.	2.1	118
78	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	3.9	117
79	Spt4 selectively regulates the expression of <i>C9orf72</i> sense and antisense mutant transcripts. Science, 2016, 353, 708-712.	6.0	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.	1.5	115
81	TREM2 in CNS homeostasis and neurodegenerative disease. Molecular Neurodegeneration, 2015, 10, 43.	4.4	115
82	Long-read sequencing across the C9orf72 â€~GGGGCCC' repeat expansion: implications for clinical use and genetic discovery efforts in human disease. Molecular Neurodegeneration, 2018, 13, 46.	4.4	111
83	Genetic and Clinical Features of Progranulin-Associated Frontotemporal Lobar Degeneration. Archives of Neurology, 2011, 68, 488.	4.9	108
84	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. Acta Neuropathologica, 2015, 130, 863-876.	3.9	104
85	<i>Fus</i> gene mutations in familial and sporadic amyotrophic lateral sclerosis. Muscle and Nerve, 2010, 42, 170-176.	1.0	101
86	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. Human Molecular Genetics, 2017, 26, 3421-3431.	1.4	101
87	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	7.1	101
88	Regional distribution of synaptic markers and APP correlate with distinct clinicopathological features in sporadic and familial Alzheimer's disease. Brain, 2014, 137, 1533-1549.	3.7	100
89	APOE ε2 is associated with increased tau pathology in primary tauopathy. Nature Communications, 2018, 9, 4388.	5.8	100
90	Lipidomic and Transcriptomic Basis of Lysosomal Dysfunction in Progranulin Deficiency. Cell Reports, 2017, 20, 2565-2574.	2.9	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.	1.8	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.	4.9	97
93	Shared genetic risk between corticobasal degeneration, progressive supranuclear palsy, and frontotemporal dementia. Acta Neuropathologica, 2017, 133, 825-837.	3.9	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.	3.9	90
95	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. Acta Neuropathologica, 2015, 130, 559-573.	3.9	89
96	A Belgian ancestral haplotype harbours a highly prevalent mutation for 17q21-linked tau-negative FTLD. Brain, 2006, 129, 841-852.	3.7	88
97	Tau pathology in frontotemporal lobar degeneration with C9ORF72 hexanucleotide repeat expansion. Acta Neuropathologica, 2013, 125, 289-302.	3.9	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.	2.1	87
99	A nonsynonymous mutation in PLCG2 reduces the risk of Alzheimer's disease, dementia with Lewy bodies and frontotemporal dementia, and increases the likelihood of longevity. Acta Neuropathologica, 2019, 138, 237-250.	3.9	87
100	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	1.5	86
101	<i>C9ORF72</i> repeat expansions in cases with previously identified pathogenic mutations. Neurology, 2013, 81, 1332-1341.	1.5	84
102	Length of normal alleles of C9ORF72 GGGGCC repeat do not influence disease phenotype. Neurobiology of Aging, 2012, 33, 2950.e5-2950.e7.	1.5	83
103	What we know about TMEM106B in neurodegeneration. Acta Neuropathologica, 2016, 132, 639-651.	3.9	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.	1.4	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.	0.8	82
106	<i>TMEM106B</i> risk variant is implicated in the pathologic presentation of Alzheimer disease. Neurology, 2012, 79, 717-718.	1.5	81
107	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
108	Hippocampal Sclerosis in the Elderly. Alzheimer Disease and Associated Disorders, 2011, 25, 364-368.	0.6	78

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109	Increased prevalence of autoimmune disease within C9 and FTD/MND cohorts. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e301.	3.1	78
110	rs5848 polymorphism and serum progranulin level. Journal of the Neurological Sciences, 2011, 300, 28-32.	0.3	77
111	The molecular genetics and neuropathology of frontotemporal lobar degeneration: recent developments. Neurogenetics, 2007, 8, 237-248.	0.7	76
112	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
113	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. Neurobiology of Aging, 2014, 35, 2421.e13-2421.e17.	1.5	74
114	Progranulin protein levels are differently regulated in plasma and CSF. Neurology, 2014, 82, 1871-1878.	1.5	70
115	Homotypic fibrillization of TMEM106B across diverse neurodegenerative diseases. Cell, 2022, 185, 1346-1355.e15.	13.5	70
116	Progranulin-associated PiB-negative logopenic primary progressive aphasia. Journal of Neurology, 2014, 261, 604-614.	1.8	69
117	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	1.5	69
118	Prosaposin is a regulator of progranulin levels and oligomerization. Nature Communications, 2016, 7, 11992.	5.8	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.	3.9	67
120	Novel mutation in MAPT exon 13 (p.N410H) causes corticobasal degeneration. Acta Neuropathologica, 2014, 127, 271-282.	3.9	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.	3.9	65
122	A Point Mutation in PDGFRB Causes Autosomal-Dominant Penttinen Syndrome. American Journal of Human Genetics, 2015, 97, 465-474.	2.6	64
123	Genetic modifiers in carriers of repeat expansions in the C9ORF72 gene. Molecular Neurodegeneration, 2014, 9, 38.	4.4	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.	4.4	61
125	Cerebellar ataxia in progressive supranuclear palsy: An autopsy study of PSP . Movement Disorders, 2016, 31, 653-662.	2.2	60
126	In vivo ¹⁸ F-AV-1451 tau PET signal in <i>MAPT</i> mutation carriers varies by expected tau isoforms. Neurology, 2018, 90, e947-e954.	1.5	60

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127	Alzheimer Disease–like Phenotype Associated With the c.154delA Mutation in Progranulin. Archives of Neurology, 2010, 67, 171-7.	4.9	59
128	Sporadic corticobasal syndrome due to FTLD-TDP. Acta Neuropathologica, 2010, 119, 365-374.	3.9	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.	4.4	58
130	SLC20A2 and THAP1 deletion in familial basal ganglia calcification with dystonia. Neurogenetics, 2014, 15, 23-30.	0.7	56
131	The neuropsychology of normal aging and preclinical Alzheimer's disease. Alzheimer's and Dementia, 2014, 10, 84-92.	0.4	55
132	Cognitive reserve and TMEM106B genotype modulate brain damage in presymptomatic frontotemporal dementia: a GENFI study. Brain, 2017, 140, 1784-1791.	3.7	55
133	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
134	Plasma Neurofilament Light for Prediction of Disease Progression in Familial Frontotemporal Lobar Degeneration. Neurology, 2021, 96, e2296-e2312.	1.5	52
135	Recent insights into the molecular genetics of dementia. Trends in Neurosciences, 2009, 32, 451-461.	4.2	51
136	Jump from Pre-mutation to Pathologic Expansion in C9orf72. American Journal of Human Genetics, 2015, 96, 962-970.	2.6	50
137	Ossified blood vessels in primary familial brain calcification elicit a neurotoxic astrocyte response. Brain, 2019, 142, 885-902.	3.7	50
138	Clinical and neuroimaging biomarkers of amyloid-negative logopenic primary progressive aphasia. Brain and Language, 2015, 142, 45-53.	0.8	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.	2.6	48
140	Altered microRNA expression in frontotemporal lobar degeneration with TDP-43 pathology caused by progranulin mutations. BMC Genomics, 2011, 12, 527.	1.2	48
141	<scp>S</scp> tudy of <i>LRRK2</i> variation in tauopathy: Progressive supranuclear palsy and corticobasal degeneration. Movement Disorders, 2017, 32, 115-123.	2.2	48
142	Poly(GP), neurofilament and grey matter deficits in <i>C9orf72</i> expansion carriers. Annals of Clinical and Translational Neurology, 2018, 5, 583-597.	1.7	48
143	C9orf72 repeat expansions in patients with ALS and FTD. Lancet Neurology, The, 2012, 11, 297-298.	4.9	46
144	Distribution and characteristics of transactive response DNA binding protein 43 kDa pathology in progressive supranuclear palsy. Movement Disorders, 2017, 32, 246-255.	2.2	46

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145	Cognitive impairment in progressive supranuclear palsy is associated with tau burden. Movement Disorders, 2017, 32, 1772-1779.	2.2	46
146	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
147	Frontotemporal lobar degeneration with TDPâ€43 proteinopathy and chromosome 9p repeat expansion in <i>C9ORF72</i> : clinicopathologic correlation. Neuropathology, 2013, 33, 122-133.	0.7	45
148	Genetic Screening and Functional Characterization of <i>PDGFRB</i> Mutations Associated with Basal Ganglia Calcification of Unknown Etiology. Human Mutation, 2014, 35, 964-971.	1.1	45
149	Loss of TMEM106B leads to myelination deficits: implications for frontotemporal dementia treatment strategies. Brain, 2020, 143, 1905-1919.	3.7	44
150	The genetics of frontotemporal lobar degeneration. Current Neurology and Neuroscience Reports, 2007, 7, 434-442.	2.0	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.	1.2	43
152	Three sib-pairs of autopsy-confirmed progressive supranuclear palsy. Parkinsonism and Related Disorders, 2015, 21, 101-105.	1.1	42
153	Loss of homeostatic microglial phenotype in CSF1R-related Leukoencephalopathy. Acta Neuropathologica Communications, 2020, 8, 72.	2.4	42
154	Genetics of Early-Onset Alzheimer Dementia. Scientific World Journal, The, 2003, 3, 497-519.	0.8	40
155	Analysis of COQ2gene in multiple system atrophy. Molecular Neurodegeneration, 2014, 9, 44.	4.4	40
156	Identical twins with the <i>C9orf72</i> repeat expansion are discordant for ALS. Neurology, 2014, 83, 1476-1478.	1.5	40
157	Hereditary diffuse leukoencephalopathy with spheroids with phenotype of primary progressive multiple sclerosis. European Journal of Neurology, 2015, 22, 328-333.	1.7	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.	3.9	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.	1.5	40
160	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. Acta Neuropathologica Communications, 2019, 7, 150.	2.4	40
161	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. Brain, 2018, 141, 2895-2907.	3.7	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.	1.7	39

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163	Association of <i>MAPT</i> Subhaplotypes With Risk of Progressive Supranuclear Palsy and Severity of Tau Pathology. JAMA Neurology, 2019, 76, 710.	4.5	39
164	A novel tau mutation, p.K317N, causes globular glial tauopathy. Acta Neuropathologica, 2015, 130, 199-214.	3.9	38
165	Clinical and neuropathological features of ALS/FTD with TIA1 mutations. Acta Neuropathologica Communications, 2017, 5, 96.	2.4	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.	1.1	37
167	Progranulin gene mutation with an unusual clinical and neuropathologic presentation. Movement Disorders, 2008, 23, 1168-1173.	2.2	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.	0.9	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.	0.8	35
170	Loss of Tmem106b exacerbates <scp>FTLD</scp> pathologies and causes motor deficits in progranulinâ€deficient mice. EMBO Reports, 2020, 21, e50197.	2.0	35
171	Network-driven plasma proteomics expose molecular changes in the Alzheimer's brain. Molecular Neurodegeneration, 2016, 11, 31.	4.4	34
172	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
173	A molecular pathology, neurobiology, biochemical, genetic and neuroimaging study of progressive apraxia of speech. Nature Communications, 2021, 12, 3452.	5.8	34
174	Ubiquitin associated protein 1 is a risk factor for frontotemporal lobar degeneration. Neurobiology of Aging, 2009, 30, 656-665.	1.5	33
175	Unaffected mosaic <i>C9orf72</i> case. Neurology, 2018, 90, e323-e331.	1.5	33
176	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. NeuroImage, 2019, 189, 645-654.	2.1	33
177	Dominant Frontotemporal Dementia Mutations in 140 Cases of Primary Progressive Aphasia and Speech Apraxia. Dementia and Geriatric Cognitive Disorders, 2015, 39, 281-286.	0.7	32
178	Sensitivity–Specificity of Tau and Amyloid β Positron Emission Tomography in Frontotemporal Lobar Degeneration. Annals of Neurology, 2020, 88, 1009-1022.	2.8	32
179	C9ORF72 repeat expansions and other FTD gene mutations in a clinical AD patient series from Mayo Clinic. American Journal of Neurodegenerative Disease, 2012, 1, 107-18.	0.1	32
180	Similarities between familial and sporadic autopsy-proven progressive supranuclear palsy. Neurology, 2013, 80, 2076-2078.	1.5	31

#	Article	IF	CITATIONS
181	Role for the microtubule-associated protein tau variant p.A152T in risk of α-synucleinopathies. Neurology, 2015, 85, 1680-1686.	1.5	31
182	Progressive amnestic dementia, hippocampal sclerosis, and mutation in C9ORF72. Acta Neuropathologica, 2013, 126, 545-554.	3.9	30
183	LRRK2 variation and dementia with Lewy bodies. Parkinsonism and Related Disorders, 2016, 31, 98-103.	1.1	30
184	TMEM106B haplotypes have distinct gene expression patterns in aged brain. Molecular Neurodegeneration, 2018, 13, 35.	4.4	30
185	Thalamo-cortical network hyperconnectivity in preclinical progranulin mutation carriers. NeuroImage: Clinical, 2019, 22, 101751.	1.4	30
186	Novel causal genes and disease modifiers. Nature Reviews Neurology, 2013, 9, 63-64.	4.9	28
187	Expanded <i>C9ORF72</i> Hexanucleotide Repeat in Depressive Pseudodementia. JAMA Neurology, 2014, 71, 775.	4.5	28
188	Differential early subcortical involvement in genetic FTD within the GENFI cohort. NeuroImage: Clinical, 2021, 30, 102646.	1.4	28
189	A Novel Tau Mutation in Exon 12, p.Q336H, Causes Hereditary Pick Disease. Journal of Neuropathology and Experimental Neurology, 2015, 74, 1042-1052.	0.9	27
190	White matter hyperintensities in progranulin-associated frontotemporal dementia: A longitudinal GENFI study. NeuroImage: Clinical, 2019, 24, 102077.	1.4	27
191	Clinical and volumetric changes with increasing functional impairment in familial frontotemporal lobar degeneration. Alzheimer's and Dementia, 2020, 16, 49-59.	0.4	27
192	A nonsense mutation in PRNP associated with clinical Alzheimer's disease. Neurobiology of Aging, 2014, 35, 2656.e13-2656.e16.	1.5	26
193	Gray matter changes in asymptomatic C9orf72 and GRN mutation carriers. NeuroImage: Clinical, 2018, 18, 591-598.	1.4	26
194	Profilin-1 mutations are rare in patients with amyotrophic lateral sclerosis and frontotemporal dementia. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 463-469.	1.1	25
195	Bromodomain inhibitors regulate the C9ORF72 locus in ALS. Experimental Neurology, 2015, 271, 241-250.	2.0	25
196	Slowly progressive dementia caused by MAPT R406W mutations: longitudinal report on a new kindred and systematic review. Alzheimer's Research and Therapy, 2018, 10, 2.	3.0	25
197	Partial Tmem106b reduction does not correct abnormalities due to progranulin haploinsufficiency. Molecular Neurodegeneration, 2018, 13, 32.	4.4	25
198	EIF2AK3 variants in Dutch patients with Alzheimer's disease. Neurobiology of Aging, 2019, 73, 229.e11-229.e18.	1.5	25

#	Article	IF	CITATIONS
199	A truncating SOD1 mutation, p.Gly141X, is associated with clinical and pathologic heterogeneity, including frontotemporal lobar degeneration. Acta Neuropathologica, 2015, 130, 145-157.	3.9	24
200	Dipeptide repeat proteins activate a heat shock response found in C9ORF72-ALS/FTLD patients. Acta Neuropathologica Communications, 2018, 6, 55.	2.4	24
201	Clinicopathologic correlations in a family with a <i>TBK1</i> mutation presenting as primary progressive aphasia and primary lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 568-575.	1.1	24
202	Gyrification abnormalities in presymptomatic <i>c9orf72</i> expansion carriers. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1005-1010.	0.9	24
203	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. Alzheimer's and Dementia, 2022, 18, 1408-1423.	0.4	24
204	Angiogenin variation and Parkinson disease. Annals of Neurology, 2012, 71, 725-727.	2.8	23
205	Revised Self-Monitoring Scale. Neurology, 2020, 94, e2384-e2395.	1.5	23
206	An adult-onset leukoencephalopathy with axonal spheroids and pigmented glia accompanied by brain calcifications. Journal of Neurology, 2013, 260, 2665-2668.	1.8	22
207	Disease and Region Specificity of Granulin Immunopositivities in Alzheimer Disease and Frontotemporal Lobar Degeneration. Journal of Neuropathology and Experimental Neurology, 2017, 76, 957-968.	0.9	22
208	Early Neuropsychological Characteristics of Progranulin Mutation Carriers. Journal of the International Neuropsychological Society, 2014, 20, 694-703.	1.2	21
209	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. Experimental Neurology, 2016, 277, 171-177.	2.0	21
210	Brain volumetric deficits in <i>MAPT</i> mutation carriers: a multisite study. Annals of Clinical and Translational Neurology, 2021, 8, 95-110.	1.7	21
211	Lewy Body Disease is a Contributor to Logopenic Progressive Aphasia Phenotype. Annals of Neurology, 2021, 89, 520-533.	2.8	21
212	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum <scp>NfL</scp> and <scp>pNfH</scp> : A Longitudinal Multicentre Study. Annals of Neurology, 2022, 91, 33-47.	2.8	21
213	Progranulin axis and recent developments in frontotemporal lobar degeneration. Alzheimer's Research and Therapy, 2012, 4, 4.	3.0	20
214	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
215	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 1782-1796.	1.7	20
216	Three VCP Mutations in Patients with Frontotemporal Dementia. Journal of Alzheimer's Disease, 2018, 65, 1139-1146.	1.2	19

#	Article	IF	CITATIONS
217	Excess of Rare Damaging TUBA4A Variants Suggests Cytoskeletal Defects in ALS. Neuron, 2014, 84, 241-243.	3.8	18
218	Frontal lobe ¹ H MR spectroscopy in asymptomatic and symptomatic <i>MAPT</i> mutation carriers. Neurology, 2019, 93, e758-e765.	1.5	18
219	Tremor in progressive supranuclear palsy. Parkinsonism and Related Disorders, 2016, 27, 93-97.	1.1	17
220	Underlying genetic variation in familial frontotemporal dementia: sequencing of 198 patients. Neurobiology of Aging, 2021, 97, 148.e9-148.e16.	1.5	17
221	Long-read targeted sequencing uncovers clinicopathological associations for <i>C9orf72</i> -linked diseases. Brain, 2021, 144, 1082-1088.	3.7	17
222	Pathologic Staging of White Matter Lesions in Adult-Onset Leukoencephalopathy/Leukodystrophy With Axonal Spheroids. Journal of Neuropathology and Experimental Neurology, 2015, 74, 233-240.	0.9	16
223	Primary familial brain calcification in the †IBGC2' kindred: All linkage roads lead to <i>SLC20A2</i> . Movement Disorders, 2016, 31, 1901-1904.	2.2	16
224	TREM2 p.R47H substitution is not associated with dementia with Lewy bodies. Neurology: Genetics, 2016, 2, e85.	0.9	16
225	FDG-PET in presymptomatic C9orf72 mutation carriers. NeuroImage: Clinical, 2021, 31, 102687.	1.4	16
226	The GGGGCC Repeat Expansion inC9ORF72in a Case with Discordant Clinical and FDG-PET Findings: PET Trumps Syndrome. Neurocase, 2014, 20, 110-120.	0.2	15
227	Brain calcifications and <i>PCDH12</i> variants. Neurology: Genetics, 2017, 3, e166.	0.9	15
228	Identification of compound heterozygous variants in <i>OPTN</i> in an ALS-FTD patient from the CReATe consortium: a case report. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 469-471.	1.1	15
229	Soluble sortilin is present in excess and positively correlates with progranulin in CSF of aging individuals. Experimental Gerontology, 2016, 84, 96-100.	1.2	14
230	Preferential Disruption of Auditory Word Representations in Primary Progressive Aphasia With the Neuropathology of FTLD-TDP Type A. Cognitive and Behavioral Neurology, 2019, 32, 46-53.	0.5	14
231	Trajectory of lobar atrophy in asymptomatic and symptomatic GRN mutation carriers: a longitudinal MRI study. Neurobiology of Aging, 2020, 88, 42-50.	1.5	14
232	Mutations in protein N-arginine methyltransferases are not the cause of FTLD-FUS. Neurobiology of Aging, 2013, 34, 2235.e11-2235.e13.	1.5	13
233	Machine learning suggests polygenic risk for cognitive dysfunction in amyotrophic lateral sclerosis. EMBO Molecular Medicine, 2021, 13, e12595.	3.3	13
234	C9orf72 repeats compromise nucleocytoplasmic transport. Nature Reviews Neurology, 2015, 11, 670-672.	4.9	12

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#	Article	IF	CITATIONS
235	Diffuse leukoencephalopathy with spheroids presenting as primary progressive aphasia. Neurology, 2015, 85, 652-653.	1.5	12
236	Abnormal expression of homeobox genes and transthyretin in <i>C9ORF72</i> expansion carriers. Neurology: Genetics, 2017, 3, e161.	0.9	12
237	GBA variation and susceptibility to multiple system atrophy. Parkinsonism and Related Disorders, 2020, 77, 64-69.	1.1	12
238	Cortical and subcortical pathological burden and neuronal loss in an autopsy series of FTLD-TDP-type C. Brain, 2022, 145, 1069-1078.	3.7	12
239	FTDPâ€17 with Pick bodyâ€like inclusions associated with a novel tau mutation, p.E372G. Brain Pathology, 2017, 27, 612-626.	2.1	11
240	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
241	Lysosomal Dysfunction and Other Pathomechanisms in FTLD: Evidence from Progranulin Genetics and Biology. Advances in Experimental Medicine and Biology, 2021, 1281, 219-242.	0.8	11
242	Identification of missing variants by combining multiple analytic pipelines. BMC Bioinformatics, 2018, 19, 139.	1.2	10
243	Revisiting the utility of TDP-43 immunoreactive (TDP-43-ir) pathology to classify FTLD-TDP subtypes. Acta Neuropathologica, 2019, 138, 167-169.	3.9	10
244	Gene Expression Imputation Across Multiple Tissue Types Provides Insight Into the Genetic Architecture of Frontotemporal Dementia and Its Clinical Subtypes. Biological Psychiatry, 2021, 89, 825-835.	0.7	10
245	Old age genetically confirmed frontotemporal lobar degeneration with TDPâ€43 has limbic predominant TDPâ€43 deposition. Neuropathology and Applied Neurobiology, 2021, 47, 1050-1059.	1.8	10
246	Assessment of Olfactory Function in MAPT-Associated Neurodegenerative Disease Reveals Odor-Identification Irreproducibility as a Non-Disease-Specific, General Characteristic of Olfactory Dysfunction. PLoS ONE, 2016, 11, e0165112.	1.1	10
247	Revealing the Mutational Spectrum in Southern Africans With Amyotrophic Lateral Sclerosis. Neurology: Genetics, 2022, 8, e654.	0.9	10
248	PRKAR1B mutations are a rare cause of FUS negative neuronal intermediate filament inclusion disease. Brain, 2015, 138, e357-e357.	3.7	9
249	FTLD-TDP With and Without GRN Mutations Cause Different Patterns of CA1 Pathology. Journal of Neuropathology and Experimental Neurology, 2019, 78, 844-853.	0.9	9
250	CSF1R mutation presenting as dementia with Lewy bodies. Neurocase, 2019, 25, 17-20.	0.2	9
251	Fulminant corticobasal degeneration: Agrypnia excitata in corticobasal syndrome. Neurology, 2016, 86, 1164-1166.	1.5	8
252	Combined Pathologies in FTLD-TDP Types A and C. Journal of Neuropathology and Experimental Neurology, 2018, 77, 405-412.	0.9	8

Rosa Rademakers

#	Article	IF	CITATIONS
253	Loss of Tmem106b leads to cerebellum Purkinje cell death and motor deficits. Brain Pathology, 2021, 31, e12945.	2.1	8
254	Disease-related cortical thinning in presymptomatic granulin mutation carriers. NeuroImage: Clinical, 2021, 29, 102540.	1.4	8
255	Adult polyglucosan body disease with <scp><i>GBE1</i></scp> haploinsufficiency and concomitant frontotemporal lobar degeneration. Neuropathology and Applied Neurobiology, 2014, 40, 778-782.	1.8	7
256	A Novel <i>GRN</i> Mutation (<i>GRN</i> c.708+6_+9delTGAG) in Frontotemporal Lobar Degeneration With TDP-43–Positive Inclusions. Journal of Neuropathology and Experimental Neurology, 2014, 73, 467-473.	0.9	7
257	<i>SLITRK2</i> , an X-linked modifier of the age at onset in <i>C9orf72</i> frontotemporal lobar degeneration. Brain, 2021, 144, 2798-2811.	3.7	7
258	A familial form of parkinsonism, dementia, and motor neuron disease: A longitudinal study. Parkinsonism and Related Disorders, 2014, 20, 1129-1134.	1.1	6
259	Shared brain transcriptomic signature in TDP-43 type A FTLD patients with or without <i>GRN</i> mutations. Brain, 2022, 145, 2472-2485.	3.7	6
260	Pathogenicity of exonic indels in fused in sarcoma in amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 424.e23-424.e24.	1.5	5
261	Clinical presentation of a patient with SLC20A2 and THAP1 deletions: Differential diagnosis of oromandibular dystonia. Parkinsonism and Related Disorders, 2015, 21, 329-331.	1.1	5
262	Neurobehavioral Characteristics of FDG-PET Defined Right-Dominant Semantic Dementia: A Longitudinal Study. Dementia and Geriatric Cognitive Disorders, 2021, 50, 17-28.	0.7	5
263	Latent trait modeling of tau neuropathology in progressive supranuclear palsy. Acta Neuropathologica, 2021, 141, 667-680.	3.9	5
264	Impact of variant-level batch effects on identification of genetic risk factors in large sequencing studies. PLoS ONE, 2021, 16, e0249305.	1.1	5
265	Unilateral neglect in a patient diagnosed with frontotemporal dementia and parkinsonism linked to chromosome 17. Acta Neuropsychiatrica, 2009, 21, 209-210.	1.0	4
266	Human Genetics as a Tool to Identify Progranulin Regulators. Journal of Molecular Neuroscience, 2011, 45, 532-537.	1.1	4
267	The presenilin 1 p.Gly206Ala mutation is a frequent cause of early-onset Alzheimer's disease in Hispanics in Florida. American Journal of Neurodegenerative Disease, 2016, 5, 94-101.	0.1	4
268	Dementia Research—A Roadmap for the Next Decade. JAMA Neurology, 2017, 74, 141.	4.5	3
269	DCTN1 variation in pathologically-confirmed PSP and CBD tauopathy. Parkinsonism and Related Disorders, 2017, 44, 151-153.	1.1	3
270	TMEM106B and myelination: rare leukodystrophy families reveal unexpected connections. Brain, 2017, 140, 3069-3080.	3.7	3

#	Article	IF	CITATIONS
271	Coexistence of Progressive Supranuclear Palsy With Pontocerebellar Atrophy and Myotonic Dystrophy Type 1. Journal of Neuropathology and Experimental Neurology, 2019, 78, 756-762.	0.9	3
272	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
273	Modifiers of LRRK2 parkinsonism: new therapeutic targets. Lancet Neurology, The, 2016, 15, 1200-1201.	4.9	2
274	Novel GRN mutation presenting as an aphasic dementia and evolving into corticobasal syndrome. Neurology: Genetics, 2017, 3, e201.	0.9	2
275	Relationships between lewy and tau pathologies in 375 consecutive nonâ€Alzheimer's olfactory bulbs. Movement Disorders, 2018, 33, 333-334.	2.2	1
276	Association of Mitochondrial DNA Genomic Variation With Risk of Pick Disease. Neurology, 2021, 96, e1755-e1760.	1.5	1
277	Frontotemporal Dementia. Blue Books of Neurology, 2010, 34, 397-416.	0.1	0
278	PL-04-01: UPDATE ON C9ORF72 RESEARCH IN FTLD AND ALS. , 2014, 10, P248-P248.		0
279	Repeat expansions in myoclonic epilepsy. Nature Genetics, 2018, 50, 477-478.	9.4	0