Ekaterina Rogaeva

List of Publications by Year in Descending Order

186 papers	22,090 citations	61 h-index	148 g-index
195 ext. papers	26,757 ext. citations	9.8 avg, IF	5.55 L-index

#	Paper	IF	Citations
186	Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort.. *Cortex*, **2022**, 150, 12-28	3.8	
185	Case of a Man with Hemichorea and Behavioral Changes: "A Red Herring".. *Movement Disorders Clinical Practice*, **2022**, 9, 501-507	2.2	0
184	Gene Expression Imputation Across Multiple Tissue Types Provides Insight Into the Genetic Architecture of Frontotemporal Dementia and Its Clinical Subtypes. *Biological Psychiatry*, **2021**, 89, 825-833	7.9	3
183	Combined epigenetic/genetic study identified an ALS age of onset modifier. *Acta Neuropathologica Communications*, **2021**, 9, 75	7.3	2
182	Whole-Genome Study of a Multigenerational Family with Essential Tremor. *Canadian Journal of Neurological Sciences*, **2021**, 1-6	1	0
181	Genomewide Association Studies of LRRK2 Modifiers of Parkinson's Disease. *Annals of Neurology*, **2021**, 90, 76-88	9.4	9
180	Novel Alzheimer Disease Risk Loci and Pathways in African American Individuals Using the African Genome Resources Panel: A Meta-analysis. *JAMA Neurology*, **2021**, 78, 102-113	17.2	32
179	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. *JAMA Network Open*, **2021**, 4, e2030194	10.4	14
178	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. *Nature Genetics*, **2021**, 53, 294-303	36.3	31
177	White matter hyperintensities in autopsy-confirmed frontotemporal lobar degeneration and Alzheimer's disease. *Alzheimer's Research and Therapy*, **2021**, 13, 129	9	4
176	MRI-visible perivascular space volumes, sleep duration and daytime dysfunction in adults with cerebrovascular disease. *Sleep Medicine*, **2021**, 83, 83-88	4.6	2
175	Amyloid-β toxicity modulates tau phosphorylation through the PAX6 signalling pathway. *Brain*, **2021**, 144, 2759-2770	11.2	4
174	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2021**, 78, 1236-1248	17.2	5
173	Differential early subcortical involvement in genetic FTD within the GENFI cohort. *NeuroImage: Clinical*, **2021**, 30, 102646	5.3	6
172	Disease-related cortical thinning in presymptomatic granulin mutation carriers. *NeuroImage: Clinical* , **2021**, 29, 102540	5.3	2
171	Early-Onset Alzheimer's Disease: What Is Missing in Research?. *Current Neurology and Neuroscience Reports*, **2021**, 21, 4	6.6	20
170	Parkinson's Disease, NOTCH3 Genetic Variants, and White Matter Hyperintensities. *Movement Disorders*, **2020**, 35, 2090-2095	7	8

169	DNA methylation age acceleration is associated with ALS age of onset and survival. *Acta Neuropathologica*, **2020**, 139, 943-946	14.3	12
168	Neuropathologic description of mutated amyotrophic lateral sclerosis. *Neurology: Genetics*, **2020**, 6, e394	3.8	3
167	Interaction of APOE4 alleles and PET tau imaging in former contact sport athletes. *NeuroImage: Clinical*, **2020**, 26, 102212	5.3	8
166	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. *Acta Neuropathologica Communications*, **2020**, 8, 5	7.3	15
165	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. *Cortex*, **2020**, 133, 384-398	3.8	7
164	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	24.1	90
163	Late-onset vs nonmendelian early-onset Alzheimer disease: A distinction without a difference?. *Neurology: Genetics*, **2020**, 6, e512	3.8	24
162	The Intersection between COVID-19, the Gene Family of ACE2 and Alzheimer's Disease. *Neuroscience Insights*, **2020**, 15, 2633105520975743	3	4
161	DNA Methylation Clocks and Their Predictive Capacity for Aging Phenotypes and Healthspan. *Neuroscience Insights*, **2020**, 15, 2633105520942221	3	22
160	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. *Annals of Neurology*, **2020**, 88, 113-122	9.4	11
159	Genetic and epigenetic study of an Alzheimer's disease family with monozygotic triplets. *Brain*, **2019**, 142, 3375-3381	11.2	6
158	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. *NeuroImage*, **2019**, 189, 645-654	7.9	18
157	Diagnostic delay in Parkinson's disease caused by PRKN mutations. *Parkinsonism and Related Disorders*, **2019**, 63, 217-220	3.6	11
156	Genetic Variation in the Ontario Neurodegenerative Disease Research Initiative. *Canadian Journal of Neurological Sciences*, **2019**, 46, 491-498	1	5
155	Cerebral perfusion changes in presymptomatic genetic frontotemporal dementia: a GENFI study. *Brain*, **2019**, 142, 1108-1120	11.2	23
154	Response to a letter to the editor. *Neurobiology of Aging*, **2019**, 78, 195-196	5.6	
153	Heritability and genetic variance of dementia with Lewy bodies. *Neurobiology of Disease*, **2019**, 127, 492-501	7.9	15
152	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. *Annals of Neurology*, **2019**, 85, 470-481	9.4	72

151	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
150	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. *Lancet Neurology, The*, **2019**, 18, 1103-1111	24.1	68
149	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
148	White matter hyperintensities in progranulin-associated frontotemporal dementia: A longitudinal GENFI study. *NeuroImage: Clinical*, **2019**, 24, 102077	5.3	13
147	Spatiotemporal analysis for detection of pre-symptomatic shape changes in neurodegenerative diseases: Initial application to the GENFI cohort. *NeuroImage*, **2019**, 188, 282-290	7.9	10
146	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. *Neurobiology of Aging*, **2019**, 77, 169-177	5.6	24
145	A comprehensive screening of copy number variability in dementia with Lewy bodies. *Neurobiology of Aging*, **2019**, 75, 223.e1-223.e10	5.6	10
144	Unaffected mosaic case: RNA foci, dipeptide proteins, but upregulated C9orf72 expression. *Neurology*, **2018**, 90, e323-e331	6.5	24
143	An APOE-independent cis-eSNP on chromosome 19q13.32 influences tau levels and late-onset Alzheimer's disease risk. *Neurobiology of Aging*, **2018**, 66, 178.e1-178.e8	5.6	8
142	Loss of CHCHD10-CHCHD2 complexes required for respiration underlies the pathogenicity of a CHCHD10 mutation in ALS. *Human Molecular Genetics*, **2018**, 27, 178-189	5.6	44
141	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
140	Clinical Reasoning: A 42-year-old man with unilateral leg weakness. *Neurology*, **2018**, 90, e1085-e1090	6.5	
139	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
138	Genetic Complexity of Early-Onset Alzheimer Disease **2018**, 29-50		6
137	Patterns of gray matter atrophy in genetic frontotemporal dementia: results from the GENFI study. *Neurobiology of Aging*, **2018**, 62, 191-196	5.6	104
136	Presymptomatic white matter integrity loss in familial frontotemporal dementia in the GENFI cohort: A cross-sectional diffusion tensor imaging study. *Annals of Clinical and Translational Neurology*, **2018**, 5, 1025-1036	5.3	29
135	Actigraphy Detects Greater Intra-Individual Variability During Gait in Non-Manifesting LRRK2 Mutation Carriers. *Journal of Parkinsons Disease*, **2018**, 8, 131-139	5.3	7
134	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. *Lancet Neurology, The*, **2018**, 17, 64-74	24.1	121

133	LRP10 in □synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1033-1034	24.1	9
132	Parkinsonism due to A53E □synuclein gene mutation: Clinical, genetic, epigenetic, and biochemical features. *Movement Disorders*, **2018**, 33, 1950-1955	7	12
131	LRP10 in □synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032-1033	24.1	9
130	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. *Brain*, **2018**, 141, 2895-2907	11.2	25
129	The relationship between brain atrophy and cognitive-behavioural symptoms in retired Canadian football players with multiple concussions. *NeuroImage: Clinical*, **2018**, 19, 551-558	5.3	24
128	Heart rate variability in leucine-rich repeat kinase 2-associated Parkinson's disease. *Movement Disorders*, **2017**, 32, 610-614	7	14
127	C9orf72 and ATXN2 repeat expansions coexist in a family with ataxia, dementia, and parkinsonism. *Movement Disorders*, **2017**, 32, 158-162	7	12
126	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. *Alzheimer's and Dementia*, **2017**, 13, 727-738	1.2	106
125	DNA methylation age-acceleration is associated with disease duration and age at onset in C9orf72 patients. *Acta Neuropathologica*, **2017**, 134, 271-279	14.3	26
124	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. *Neurobiology of Aging*, **2017**, 57, 247.e9-247.e13	5.6	54
123	White matter hyperintensities are seen only in mutation carriers in the GENFI cohort. *NeuroImage: Clinical*, **2017**, 15, 171-180	5.3	43
122	Collagenosis of the Deep Medullary Veins: An Underrecognized Pathologic Correlate of White Matter Hyperintensities and Periventricular Infarction?. *Journal of Neuropathology and Experimental Neurology*, **2017**, 76, 299-312	3.1	56
121	Genetic analysis of CHCHD2 and CHCHD10 in Italian patients with Parkinson's disease. *Neurobiology of Aging*, **2017**, 53, 193.e7-193.e8	5.6	7
120	Ultra-rare mutations in segregate in Caribbean Hispanic families with Alzheimer disease. *Neurology: Genetics*, **2017**, 3, e178	3.8	5
119	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. *Nature Genetics*, **2017**, 49, 1373-1384	36.3	508
118	Dysregulation of chromatin remodelling complexes in amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2017**, 26, 4142-4152	5.6	19
117	Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. *Neurobiology of Aging*, **2017**, 49, 214.e13-214.e15	5.6	10
116	Time-course global proteome analyses reveal an inverse correlation between A□burden and immunoglobulin M levels in the APPNL-F mouse model of Alzheimer disease. *PLoS ONE*, **2017**, 12, e0182844	3.7	3

115	A novel Alzheimer disease locus located near the gene encoding tau protein. *Molecular Psychiatry*, **2016**, 21, 108-17	15.1	175
114	Marked Differences in C9orf72 Methylation Status and Isoform Expression between C9/ALS Human Embryonic and Induced Pluripotent Stem Cells. *Stem Cell Reports*, **2016**, 7, 927-940	8	14
113	Mutation analysis of CHCHD2 in Canadian patients with familial Parkinson's disease. *Neurobiology of Aging*, **2016**, 38, 217.e7-217.e8	5.6	15
112	Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. *Neurobiology of Aging*, **2016**, 38, 214.e7-214.e10	5.6	49
111	MTHFSD and DDX58 are novel RNA-binding proteins abnormally regulated in amyotrophic lateral sclerosis. *Brain*, **2016**, 139, 86-100	11.2	28
110	Drug Repositioning for Alzheimer's Disease Based on Systematic 'omics' Data Mining. *PLoS ONE*, **2016**, 11, e0168812	3.7	62
109	Genetic and epigenetic study of ALS-discordant identical twins with double mutations in SOD1 and ARHGEF28. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 1268-1270	5.5	25
108	Genetic Variations in ABCA7 Can Increase Secreted Levels of Amyloid-β40 and Amyloid-β42 Peptides and ABCA7 Transcription in Cell Culture Models. *Journal of Alzheimer's Disease*, **2016**, 53, 875-92	4.3	13
107	Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. *Neurobiology of Aging*, **2016**, 42, 217.e7-217.e13	5.6	24
106	C9orf72 isoforms in Amyotrophic Lateral Sclerosis and Frontotemporal Lobar Degeneration. *Brain Research*, **2016**, 1647, 43-49	3.7	31
105	Does BDNF Val66Met contribute to preclinical Alzheimer's disease?. *Brain*, **2016**, 139, 2586-2589	11.2	6
104	Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease. *Annals of Neurology*, **2016**, 79, 739-747	9.4	42
103	The C9orf72 repeat expansion itself is methylated in ALS and FTLD patients. *Acta Neuropathologica*, **2015**, 129, 715-27	14.3	101
102	Drug repositioning for diabetes based on 'omics' data mining. *PLoS ONE*, **2015**, 10, e0126082	3.7	49
101	Mutation analysis of C9orf72 in patients with corticobasal syndrome. *Neurobiology of Aging*, **2015**, 36, 2905.e1-5	5.6	11
100	Inbreeding among Caribbean Hispanics from the Dominican Republic and its effects on risk of Alzheimer disease. *Genetics in Medicine*, **2015**, 17, 639-43	8.1	16
99	Low molecular weight species of TDP-43 generated by abnormal splicing form inclusions in amyotrophic lateral sclerosis and result in motor neuron death. *Acta Neuropathologica*, **2015**, 130, 49-61	14.3	49
98	Mutation analysis of CHCHD10 in different neurodegenerative diseases. *Brain*, **2015**, 138, e380	11.2	67

97	Association of Long Runs of Homozygosity With Alzheimer Disease Among African American Individuals. *JAMA Neurology*, **2015**, 72, 1313-23	17.2	27
96	Mutation analysis of patients with neurodegenerative disorders using NeuroX array. *Neurobiology of Aging*, **2015**, 36, 545.e9-14	5.6	24
95	Coding mutations in SORL1 and Alzheimer disease. *Annals of Neurology*, **2015**, 77, 215-27	9.4	125
94	F-box/LRR-repeat protein 7 is genetically associated with Alzheimer's disease. *Annals of Clinical and Translational Neurology*, **2015**, 2, 810-20	5.3	34
93	Rare coding mutations identified by sequencing of Alzheimer disease genome-wide association studies loci. *Annals of Neurology*, **2015**, 78, 487-98	9.4	102
92	Isoform-specific antibodies reveal distinct subcellular localizations of C9orf72 in amyotrophic lateral sclerosis. *Annals of Neurology*, **2015**, 78, 568-83	9.4	100
91	Jump from pre-mutation to pathologic expansion in C9orf72. *American Journal of Human Genetics*, **2015**, 96, 962-70	11	41
90	Rarity of the Alzheimer disease-protective APP A673T variant in the United States. *JAMA Neurology* , **2015**, 72, 209-16	17.2	31
89	Presymptomatic cognitive and neuroanatomical changes in genetic frontotemporal dementia in the Genetic Frontotemporal dementia Initiative (GENFI) study: a cross-sectional analysis. *Lancet Neurology, The*, **2015**, 14, 253-62	24.1	328
88	The Prion Protein Controls Polysialylation of Neural Cell Adhesion Molecule 1 during Cellular Morphogenesis. *PLoS ONE*, **2015**, 10, e0133741	3.7	30
87	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. *Nature Neuroscience*, **2014**, 17, 664-666	25.5	319
86	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. *Nature*, **2014** , 505, 550-554	50.4	345
85	Motor neuron disease and frontotemporal dementia: sometimes related, sometimes not. *Experimental Neurology*, **2014**, 262 Pt B, 75-83	5.7	63
84	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. *Brain* , **2014**, 137, e311	11.2	89
83	Identical twins with the C9orf72 repeat expansion are discordant for ALS. *Neurology*, **2014**, 83, 1476-8	6.5	39
82	Frontotemporal dementia and its subtypes: a genome-wide association study. *Lancet Neurology, The*, **2014**, 13, 686-99	24.1	207
81	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. *Human Molecular Genetics*, **2014**, 23, 6139-46	5.6	152
80	Hypermethylation of the CpG-island near the C9orf72 G□□-repeat expansion in FTLD patients. *Human Molecular Genetics*, **2014**, 23, 5630-7	5.6	68

79	Effects of multiple genetic loci on age at onset in late-onset Alzheimer disease: a genome-wide association study. *JAMA Neurology*, **2014**, 71, 1394-404	17.2	129
78	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
77	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
76	Evidence of recessive Alzheimer disease loci in a Caribbean Hispanic data set: genome-wide survey of runs of homozygosity. *JAMA Neurology*, **2013**, 70, 1261-7	17.2	33
75	TREM2 variants in Alzheimer's disease. *New England Journal of Medicine*, **2013**, 368, 117-27	59.2	1805
74	Autosomal dominant frontotemporal lobar degeneration due to the C9ORF72 hexanucleotide repeat expansion: late-onset psychotic clinical presentation. *Biological Psychiatry*, **2013**, 74, 384-91	7.9	94
73	Clathrin adaptor CALM/PICALM is associated with neurofibrillary tangles and is cleaved in Alzheimer's brains. *Acta Neuropathologica*, **2013**, 125, 861-78	14.3	88
72	Hypermethylation of the CpG island near the G4C2 repeat in ALS with a C9orf72 expansion. *American Journal of Human Genetics*, **2013**, 92, 981-9	11	211
71	Association between early-onset Parkinson disease and 22q11.2 deletion syndrome: identification of a novel genetic form of Parkinson disease and its clinical implications. *JAMA Neurology*, **2013**, 70, 1359-66	17.2	107
70	Variant Alzheimer's disease with spastic paraparesis and supranuclear gaze palsy. *Canadian Journal of Neurological Sciences*, **2013**, 40, 249-51	1	5
69	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. *JAMA Neurology*, **2013**, 70, 727-35	17.2	285
68	A multi-centre clinico-genetic analysis of the VPS35 gene in Parkinson disease indicates reduced penetrance for disease-associated variants. *Journal of Medical Genetics*, **2012**, 49, 721-6	5.8	78
67	Investigation of c9orf72 in 4 neurodegenerative disorders. *Archives of Neurology*, **2012**, 69, 1583-90		83
66	LIV-1 ZIP ectodomain shedding in prion-infected mice resembles cellular response to transition metal starvation. *Journal of Molecular Biology*, **2012**, 422, 556-574	6.5	29
65	Fc receptor polymorphisms do not predict response to intravenous immunoglobulin in myasthenia gravis. *Journal of Clinical Neuromuscular Disease*, **2012**, 14, 1-6	1.1	3
64	RNA targets of TDP-43 identified by UV-CLIP are deregulated in ALS. *Molecular and Cellular Neurosciences*, **2011**, 47, 167-80	4.8	130
63	A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9p21-linked ALS-FTD. *Neuron*, **2011**, 72, 257-68	13.9	3018
62	Genetics and genomics of late-onset Alzheimer's disease and its endophenotypes. *International Journal of Alzheimers Disease*, **2011**, 2011, 284728	3.7	7

61	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. *Nature Genetics*, **2011**, 43, 436-41	36.3	1367
60	SORCS1 alters amyloid precursor protein processing and variants may increase Alzheimer's disease risk. *Annals of Neurology*, **2011**, 69, 47-64	9.4	79
59	Identification of novel loci for Alzheimer disease and replication of CLU, PICALM, and BIN1 in Caribbean Hispanic individuals. *Archives of Neurology*, **2011**, 68, 320-8		135
58	Meta-analysis of the association between variants in SORL1 and Alzheimer disease. *Archives of Neurology*, **2011**, 68, 99-106		135
57	Comprehensive mutational analysis of LRRK2 reveals variants supporting association with autosomal dominant Parkinson's disease. *Journal of Human Genetics*, **2011**, 56, 671-5	4.3	9
56	Intra-familial clinical heterogeneity due to FTLD-U with TDP-43 proteinopathy caused by a novel deletion in progranulin gene (PGRN). *Journal of Alzheimers Disease*, **2010**, 22, 1123-33	4.3	16
55	Distinct biochemical signatures characterize peripherin isoform expression in both traumatic neuronal injury and motor neuron disease. *Journal of Neurochemistry*, **2010**, 114, 1177-92	6	13
54	Meta-analysis confirms CR1, CLU, and PICALM as alzheimer disease risk loci and reveals interactions with APOE genotypes. *Archives of Neurology*, **2010**, 67, 1473-84		330
53	LRRK2 and Parkin mutations in a family with parkinsonism-Lack of genotype-phenotype correlation. *Neurobiology of Aging*, **2010**, 31, 721-2	5.6	6
52	Genetic screening of Alzheimer's disease genes in Iberian and African samples yields novel mutations in presenilins and APP. *Neurobiology of Aging*, **2010**, 31, 725-31	5.6	162
51	A novel PS1 gene mutation in a large Aboriginal kindred. *Canadian Journal of Neurological Sciences*, **2010**, 37, 359-64	1	9
50	Amyotrophic lateral sclerosis is a non-amyloid disease in which extensive misfolding of SOD1 is unique to the familial form. *Acta Neuropathologica*, **2010**, 119, 335-44	14.3	147
49	Olfactory heterogeneity in LRRK2 related Parkinsonism. *Movement Disorders*, **2010**, 25, 2879-83	7	28
48	Long-term statin therapy and CSF cholesterol levels: implications for Alzheimer's disease. *Dementia and Geriatric Cognitive Disorders*, **2009**, 27, 519-24	2.6	21
47	Unilateral pallidotomy in a patient with parkinsonism and G2019S LRRK2 mutation. *Movement Disorders*, **2009**, 24, 791-2; author reply 792	7	3
46	Frequent missense and insertion/deletion polymorphisms in the ovine Shadoo gene parallel species-specific variation in PrP. *PLoS ONE*, **2009**, 4, e6538	3.7	11
45	Genetic studies of GRN and IFT74 in amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2008**, 29, 1279-82	5.6	8
44	Microbleed topography, leukoaraiosis, and cognition in probable Alzheimer disease from the Sunnybrook dementia study. *Archives of Neurology*, **2008**, 65, 790-5		205

43	Age-at-onset linkage analysis in Caribbean Hispanics with familial late-onset Alzheimer's disease. *Neurogenetics*, **2008**, 9, 51-60	3	24
42	Further examination of the candidate genes in chromosome 12p13 locus for late-onset Alzheimer disease. *Neurogenetics*, **2008**, 9, 127-38	3	23
41	The G2019S LRRK2 mutation in Brazilian patients with Parkinson's disease: phenotype in monozygotic twins. *Movement Disorders*, **2008**, 23, 290-4	7	14
40	The neuronal sortilin-related receptor SORL1 is genetically associated with Alzheimer disease. *Nature Genetics*, **2007**, 39, 168-77	36.3	888
39	Deciphering the role of heterozygous mutations in genes associated with parkinsonism. *Lancet Neurology, The*, **2007**, 6, 652-62	24.1	242
38	A novel mutation in the SPG3A gene (atlastin) in hereditary spastic paraplegia. *Journal of Neurology* , **2007**, 254, 972-4	5.5	4
37	The association between genetic variants in SORL1 and Alzheimer disease in an urban, multiethnic, community-based cohort. *Archives of Neurology*, **2007**, 64, 501-6		126
36	Statins differentially affect amyloid precursor protein metabolism in presymptomatic PS1 and non-PS1 subjects. *Archives of Neurology*, **2007**, 64, 1672-3		13
35	Association studies between the plasmin genes and late-onset Alzheimer's disease. *Neurobiology of Aging*, **2007**, 28, 1041-3	5.6	9
34	Clinical and genetic study of a Brazilian family with spastic paraplegia (SPG6 locus). *Movement Disorders*, **2006**, 21, 279-81	7	17
33	Homozygous and heterozygous PINK1 mutations: considerations for diagnosis and care of Parkinson's disease patients. *Movement Disorders*, **2006**, 21, 875-9	7	23
32	Childhood onset in familial prion disease with a novel mutation in the PRNP gene. *Archives of Neurology*, **2006**, 63, 1016-21		18
31	Genetic variability in CHMP2B and frontotemporal dementia. *Neurodegenerative Diseases*, **2006**, 3, 129-32	2.3	42
30	T313M PINK1 mutation in an extended highly consanguineous Saudi family with early-onset Parkinson disease. *Archives of Neurology*, **2006**, 63, 1483-5		18
29	Association studies of cholesterol metabolism genes (CH25H, ABCA1 and CH24H) in Alzheimer's disease. *Neuroscience Letters*, **2006**, 391, 142-6	3.3	58
28	Expanded genomewide scan implicates a novel locus at 3q28 among Caribbean hispanics with familial Alzheimer disease. *Archives of Neurology*, **2006**, 63, 1591-8		32
27	Genetic complexity of Alzheimer's disease: successes and challenges. *Journal of Alzheimer's Disease*, **2006**, 9, 381-7	4.3	46
26	Novel splicing mutation in the progranulin gene causing familial corticobasal syndrome. *Brain*, **2006** , 129, 3115-23	11.2	162

25	TMP21 is a presenilin complex component that modulates gamma-secretase but not epsilon-secretase activity. *Nature*, **2006**, 440, 1208-12	50.4	260
24	Analysis of the glucocerebrosidase gene in Parkinson's disease. *Movement Disorders*, **2005**, 20, 367-70	7	93
23	Wild-type PINK1 prevents basal and induced neuronal apoptosis, a protective effect abrogated by Parkinson disease-related mutations. *Journal of Biological Chemistry*, **2005**, 280, 34025-32	5.4	249
22	Conversion to dementia among two groups with cognitive impairment. A preliminary report. *Dementia and Geriatric Cognitive Disorders*, **2004**, 18, 307-13	2.6	54
21	Analysis of the PINK1 gene in a large cohort of cases with Parkinson disease. *Archives of Neurology*, **2004**, 61, 1898-904		131
20	Clinical findings in a large family with a parkin ex3delta40 mutation. *Archives of Neurology*, **2004**, 61, 701-4		23
19	Lack of association between Alzheimer's disease and the promoter region polymorphisms of the nicastrin gene. *Neuroscience Letters*, **2004**, 363, 49-53	3.3	6
18	No evidence for tau duplications in frontal temporal dementia families showing genetic linkage to the tau locus in which tau mutations have not been found. *Neuroscience Letters*, **2004**, 363, 99-101	3.3	7
17	Genetic association study of PINK1 coding polymorphisms in Parkinson's disease. *Neuroscience Letters*, **2004**, 372, 226-9	3.3	28
16	Benign hereditary chorea: clinical, genetic, and pathological findings. *Annals of Neurology*, **2003**, 54, 244-7	9.4	81
15	Early-onset Parkinson's disease caused by a compound heterozygous DJ-1 mutation. *Annals of Neurology*, **2003**, 54, 271-4	9.4	202
14	Brain levels of CDK5 activator p25 are not increased in Alzheimer's or other neurodegenerative diseases with neurofibrillary tangles. *Journal of Neurochemistry*, **2003**, 86, 572-81	6	74
13	Absence of association between Alzheimer disease and the regulatory region polymorphism of the PS2 gene in an Italian population. *Neuroscience Letters*, **2003**, 343, 210-2	3.3	10
12	The solved and unsolved mysteries of the genetics of early-onset Alzheimer's disease. *NeuroMolecular Medicine*, **2002**, 2, 1-10	4.6	63
11	Nicastrin binds to membrane-tethered Notch. *Nature Cell Biology*, **2001**, 3, 751-4	23.4	118
10	Genetic markers in the diagnosis of Alzheimer's disease. *Journal of Alzheimer's Disease*, **2001**, 3, 293-304	4.3	7
9	Molecular genetics of Alzheimer's disease: the role of beta-amyloid and the presenilins. *Current Opinion in Neurology*, **2000**, 13, 377-84	7.1	60
8	Mutation of the conserved N-terminal cysteine (Cys92) of human presenilin 1 causes increased A beta42 secretion in mammalian cells but impaired Notch/lin-12 signalling in C. elegans. *NeuroReport* , **2000**, 11, 3227-30	1.7	29

7	Nicastrin modulates presenilin-mediated notch/glp-1 signal transduction and betaAPP processing. *Nature*, **2000**, 407, 48-54	50.4	829
6	Comparison of clinical and pathological phenotypes in two ethnically and geographically unrelated pedigrees segregating an equivalent presenilin 1 mutation. *Journal of Neuropsychiatry and Clinical Neurosciences*, **2000**, 12, 359-63	2.7	2
5	Novel presenilin 1 mutations associated with early onset of dementia in a family with both early-onset and late-onset Alzheimer disease. *Archives of Neurology*, **2000**, 57, 1454-7		24
4	Carboxyl-terminal fragments of Alzheimer beta-amyloid precursor protein accumulate in restricted and unpredicted intracellular compartments in presenilin 1-deficient cells. *Journal of Biological Chemistry*, **2000**, 275, 36794-802	5.4	65
3	A presenilin-1 Thr116Asn substitution in a family with early-onset Alzheimer's disease. *NeuroReport* , **1999**, 10, 2255-60	1.7	25
2	Amyloid-beta-protein isoforms in brain of subjects with PS1-linked, beta APP-linked and sporadic Alzheimer disease. *Molecular Brain Research*, **1998**, 56, 178-85		23
1	Evidence for an Alzheimer disease susceptibility locus on chromosome 12 and for further locus heterogeneity. *JAMA - Journal of the American Medical Association*, **1998**, 280, 614-8	27.4	107