

Ekaterina Rogaeva

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9p21-linked ALS-FTD. <i>Neuron</i> , 2011 , 72, 257-68	13.9	3018
185	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. <i>Nature Genetics</i> , 2013 , 45, 1452-8	36.3	2714
184	TREM2 variants in Alzheimer's disease. <i>New England Journal of Medicine</i> , 2013 , 368, 117-27	59.2	1805
183	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. <i>Nature Genetics</i> , 2011 , 43, 436-41	36.3	1367
182	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A β tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019 , 51, 414-430	36.3	917
181	The neuronal sortilin-related receptor SORL1 is genetically associated with Alzheimer disease. <i>Nature Genetics</i> , 2007 , 39, 168-77	36.3	888
180	Nicastrin modulates presenilin-mediated notch/glp-1 signal transduction and betaAPP processing. <i>Nature</i> , 2000 , 407, 48-54	50.4	829
179	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. <i>Nature Genetics</i> , 2017 , 49, 1373-1384	36.3	508
178	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. <i>Nature</i> , 2014 , 505, 550-554	50.4	345
177	Meta-analysis confirms CR1, CLU, and PICALM as alzheimer disease risk loci and reveals interactions with APOE genotypes. <i>Archives of Neurology</i> , 2010 , 67, 1473-84		330
176	Presymptomatic cognitive and neuroanatomical changes in genetic frontotemporal dementia in the Genetic Frontotemporal dementia Initiative (GENFI) study: a cross-sectional analysis. <i>Lancet Neurology</i> , 2015 , 14, 253-62	24.1	328
175	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. <i>Nature Neuroscience</i> , 2014 , 17, 664-666	25.5	319
174	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018 , 97, 1268-1283.e6	13.9	296
173	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. <i>JAMA Neurology</i> , 2013 , 70, 727-35	17.2	285
172	TMP21 is a presenilin complex component that modulates gamma-secretase but not epsilon-secretase activity. <i>Nature</i> , 2006 , 440, 1208-12	50.4	260
171	Wild-type PINK1 prevents basal and induced neuronal apoptosis, a protective effect abrogated by Parkinson disease-related mutations. <i>Journal of Biological Chemistry</i> , 2005 , 280, 34025-32	5.4	249
170	Deciphering the role of heterozygous mutations in genes associated with parkinsonism. <i>Lancet Neurology</i> , 2007 , 6, 652-62	24.1	242

169	Hypermethylation of the CpG island near the G4C2 repeat in ALS with a C9orf72 expansion. <i>American Journal of Human Genetics</i> , 2013 , 92, 981-9	11	211
168	Frontotemporal dementia and its subtypes: a genome-wide association study. <i>Lancet Neurology, The</i> , 2014 , 13, 686-99	24.1	207
167	Microbleed topography, leukoaraiosis, and cognition in probable Alzheimer disease from the Sunnybrook dementia study. <i>Archives of Neurology</i> , 2008 , 65, 790-5		205
166	Early-onset Parkinson's disease caused by a compound heterozygous DJ-1 mutation. <i>Annals of Neurology</i> , 2003 , 54, 271-4	9.4	202
165	A novel Alzheimer disease locus located near the gene encoding tau protein. <i>Molecular Psychiatry</i> , 2016 , 21, 108-17	15.1	175
164	Genetic screening of Alzheimer's disease genes in Iberian and African samples yields novel mutations in presenilins and APP. <i>Neurobiology of Aging</i> , 2010 , 31, 725-31	5.6	162
163	Novel splicing mutation in the progranulin gene causing familial corticobasal syndrome. <i>Brain</i> , 2006 , 129, 3115-23	11.2	162
162	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. <i>Human Molecular Genetics</i> , 2014 , 23, 6139-46	5.6	152
161	Amyotrophic lateral sclerosis is a non-amyloid disease in which extensive misfolding of SOD1 is unique to the familial form. <i>Acta Neuropathologica</i> , 2010 , 119, 335-44	14.3	147
160	Identification of novel loci for Alzheimer disease and replication of CLU, PICALM, and BIN1 in Caribbean Hispanic individuals. <i>Archives of Neurology</i> , 2011 , 68, 320-8		135
159	Meta-analysis of the association between variants in SORL1 and Alzheimer disease. <i>Archives of Neurology</i> , 2011 , 68, 99-106		135
158	Analysis of the PINK1 gene in a large cohort of cases with Parkinson disease. <i>Archives of Neurology</i> , 2004 , 61, 1898-904		131
157	RNA targets of TDP-43 identified by UV-CLIP are deregulated in ALS. <i>Molecular and Cellular Neurosciences</i> , 2011 , 47, 167-80	4.8	130
156	Effects of multiple genetic loci on age at onset in late-onset Alzheimer disease: a genome-wide association study. <i>JAMA Neurology</i> , 2014 , 71, 1394-404	17.2	129
155	The association between genetic variants in SORL1 and Alzheimer disease in an urban, multiethnic, community-based cohort. <i>Archives of Neurology</i> , 2007 , 64, 501-6		126
154	Coding mutations in SORL1 and Alzheimer disease. <i>Annals of Neurology</i> , 2015 , 77, 215-27	9.4	125
153	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. <i>Lancet Neurology, The</i> , 2018 , 17, 64-74	24.1	121
152	Nicastrin binds to membrane-tethered Notch. <i>Nature Cell Biology</i> , 2001 , 3, 751-4	23.4	118

151	Association between early-onset Parkinson disease and 22q11.2 deletion syndrome: identification of a novel genetic form of Parkinson disease and its clinical implications. <i>JAMA Neurology</i> , 2013 , 70, 1359-66	17.2	107
150	Evidence for an Alzheimer disease susceptibility locus on chromosome 12 and for further locus heterogeneity. <i>JAMA - Journal of the American Medical Association</i> , 1998 , 280, 614-8	27.4	107
149	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. <i>Alzheimers and Dementia</i> , 2017 , 13, 727-738	1.2	106
148	Patterns of gray matter atrophy in genetic frontotemporal dementia: results from the GENFI study. <i>Neurobiology of Aging</i> , 2018 , 62, 191-196	5.6	104
147	Rare coding mutations identified by sequencing of Alzheimer disease genome-wide association studies loci. <i>Annals of Neurology</i> , 2015 , 78, 487-98	9.4	102
146	The C9orf72 repeat expansion itself is methylated in ALS and FTLD patients. <i>Acta Neuropathologica</i> , 2015 , 129, 715-27	14.3	101
145	Isoform-specific antibodies reveal distinct subcellular localizations of C9orf72 in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2015 , 78, 568-83	9.4	100
144	Autosomal dominant frontotemporal lobar degeneration due to the C9ORF72 hexanucleotide repeat expansion: late-onset psychotic clinical presentation. <i>Biological Psychiatry</i> , 2013 , 74, 384-91	7.9	94
143	Analysis of the glucocerebrosidase gene in Parkinson's disease. <i>Movement Disorders</i> , 2005 , 20, 367-70	7	93
142	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. <i>PLoS ONE</i> , 2014 , 9, e94661	3.7	90
141	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. <i>Lancet Neurology</i> , 2020 , 19, 145-156	24.1	90
140	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. <i>Brain</i> , 2014 , 137, e311	11.2	89
139	Clathrin adaptor CALM/PICALM is associated with neurofibrillary tangles and is cleaved in Alzheimer's brains. <i>Acta Neuropathologica</i> , 2013 , 125, 861-78	14.3	88
138	Investigation of c9orf72 in 4 neurodegenerative disorders. <i>Archives of Neurology</i> , 2012 , 69, 1583-90		83
137	Benign hereditary chorea: clinical, genetic, and pathological findings. <i>Annals of Neurology</i> , 2003 , 54, 244-54	7.4	81
136	SORCS1 alters amyloid precursor protein processing and variants may increase Alzheimer's disease risk. <i>Annals of Neurology</i> , 2011 , 69, 47-64	9.4	79
135	A multi-centre clinico-genetic analysis of the VPS35 gene in Parkinson disease indicates reduced penetrance for disease-associated variants. <i>Journal of Medical Genetics</i> , 2012 , 49, 721-6	5.8	78
134	Brain levels of CDK5 activator p25 are not increased in Alzheimer's or other neurodegenerative diseases with neurofibrillary tangles. <i>Journal of Neurochemistry</i> , 2003 , 86, 572-81	6	74

133	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019 , 85, 470-481	9.4	72
132	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. <i>Lancet Neurology</i> , 2019 , 18, 1103-1111	24.1	68
131	Hypermethylation of the CpG-island near the C9orf72 GGGRepeat expansion in FTLD patients. <i>Human Molecular Genetics</i> , 2014 , 23, 5630-7	5.6	68
130	Mutation analysis of CHCHD10 in different neurodegenerative diseases. <i>Brain</i> , 2015 , 138, e380	11.2	67
129	Carboxyl-terminal fragments of Alzheimer beta-amyloid precursor protein accumulate in restricted and unpredicted intracellular compartments in presenilin 1-deficient cells. <i>Journal of Biological Chemistry</i> , 2000 , 275, 36794-802	5.4	65
128	Motor neuron disease and frontotemporal dementia: sometimes related, sometimes not. <i>Experimental Neurology</i> , 2014 , 262 Pt B, 75-83	5.7	63
127	The solved and unsolved mysteries of the genetics of early-onset Alzheimer's disease. <i>NeuroMolecular Medicine</i> , 2002 , 2, 1-10	4.6	63
126	Drug Repositioning for Alzheimer's Disease Based on Systematic 'omics' Data Mining. <i>PLoS ONE</i> , 2016 , 11, e0168812	3.7	62
125	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. <i>Lancet Neurology</i> , 2018 , 17, 548-558	24.1	60
124	Molecular genetics of Alzheimer's disease: the role of beta-amyloid and the presenilins. <i>Current Opinion in Neurology</i> , 2000 , 13, 377-84	7.1	60
123	Association studies of cholesterol metabolism genes (CH25H, ABCA1 and CH24H) in Alzheimer's disease. <i>Neuroscience Letters</i> , 2006 , 391, 142-6	3.3	58
122	Collagenosis of the Deep Medullary Veins: An Underrecognized Pathologic Correlate of White Matter Hyperintensities and Periventricular Infarction?. <i>Journal of Neuropathology and Experimental Neurology</i> , 2017 , 76, 299-312	3.1	56
121	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017 , 57, 247.e9-247.e13	5.6	54
120	Conversion to dementia among two groups with cognitive impairment. A preliminary report. <i>Dementia and Geriatric Cognitive Disorders</i> , 2004 , 18, 307-13	2.6	54
119	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. <i>Acta Neuropathologica</i> , 2019 , 137, 879-899	14.3	50
118	Drug repositioning for diabetes based on 'omics' data mining. <i>PLoS ONE</i> , 2015 , 10, e0126082	3.7	49
117	Low molecular weight species of TDP-43 generated by abnormal splicing form inclusions in amyotrophic lateral sclerosis and result in motor neuron death. <i>Acta Neuropathologica</i> , 2015 , 130, 49-61	14.3	49
116	Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. <i>Neurobiology of Aging</i> , 2016 , 38, 214.e7-214.e10	5.6	49

115	Genetic complexity of Alzheimer's disease: successes and challenges. <i>Journal of Alzheimers Disease</i> , 2006 , 9, 381-7	4.3	46
114	Loss of CHCHD10-CHCHD2 complexes required for respiration underlies the pathogenicity of a CHCHD10 mutation in ALS. <i>Human Molecular Genetics</i> , 2018 , 27, 178-189	5.6	44
113	White matter hyperintensities are seen only in mutation carriers in the GENFI cohort. <i>NeuroImage: Clinical</i> , 2017 , 15, 171-180	5.3	43
112	Genetic variability in CHMP2B and frontotemporal dementia. <i>Neurodegenerative Diseases</i> , 2006 , 3, 129-33.3		42
111	Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease. <i>Annals of Neurology</i> , 2016 , 79, 739-747	9.4	42
110	Jump from pre-mutation to pathologic expansion in C9orf72. <i>American Journal of Human Genetics</i> , 2015 , 96, 962-70	11	41
109	Identical twins with the C9orf72 repeat expansion are discordant for ALS. <i>Neurology</i> , 2014 , 83, 1476-8	6.5	39
108	F-box/LRR-repeat protein 7 is genetically associated with Alzheimer's disease. <i>Annals of Clinical and Translational Neurology</i> , 2015 , 2, 810-20	5.3	34
107	Evidence of recessive Alzheimer disease loci in a Caribbean Hispanic data set: genome-wide survey of runs of homozygosity. <i>JAMA Neurology</i> , 2013 , 70, 1261-7	17.2	33
106	Expanded genomewide scan implicates a novel locus at 3q28 among Caribbean hispanics with familial Alzheimer disease. <i>Archives of Neurology</i> , 2006 , 63, 1591-8		32
105	Novel Alzheimer Disease Risk Loci and Pathways in African American Individuals Using the African Genome Resources Panel: A Meta-analysis. <i>JAMA Neurology</i> , 2021 , 78, 102-113	17.2	32
104	Rarity of the Alzheimer disease-protective APP A673T variant in the United States. <i>JAMA Neurology</i> , 2015 , 72, 209-16	17.2	31
103	C9orf72 isoforms in Amyotrophic Lateral Sclerosis and Frontotemporal Lobar Degeneration. <i>Brain Research</i> , 2016 , 1647, 43-49	3.7	31
102	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. <i>Nature Genetics</i> , 2021 , 53, 294-303	36.3	31
101	The Prion Protein Controls Polysialylation of Neural Cell Adhesion Molecule 1 during Cellular Morphogenesis. <i>PLoS ONE</i> , 2015 , 10, e0133741	3.7	30
100	Presymptomatic white matter integrity loss in familial frontotemporal dementia in the GENFI cohort: A cross-sectional diffusion tensor imaging study. <i>Annals of Clinical and Translational Neurology</i> , 2018 , 5, 1025-1036	5.3	29
99	LIV-1 ZIP ectodomain shedding in prion-infected mice resembles cellular response to transition metal starvation. <i>Journal of Molecular Biology</i> , 2012 , 422, 556-574	6.5	29
98	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 <i>C. elegans</i> . <i>NeuroReport</i> , 2000 , 11, 3227-30	1.7	29

97	MTHFSD and DDX58 are novel RNA-binding proteins abnormally regulated in amyotrophic lateral sclerosis. <i>Brain</i> , 2016 , 139, 86-100	11.2	28
96	Olfactory heterogeneity in LRRK2 related Parkinsonism. <i>Movement Disorders</i> , 2010 , 25, 2879-83	7	28
95	Genetic association study of PINK1 coding polymorphisms in Parkinson's disease. <i>Neuroscience Letters</i> , 2004 , 372, 226-9	3.3	28
94	Association of Long Runs of Homozygosity With Alzheimer Disease Among African American Individuals. <i>JAMA Neurology</i> , 2015 , 72, 1313-23	17.2	27
93	DNA methylation age-acceleration is associated with disease duration and age at onset in C9orf72 patients. <i>Acta Neuropathologica</i> , 2017 , 134, 271-279	14.3	26
92	A presenilin-1 Thr116Asn substitution in a family with early-onset Alzheimer's disease. <i>NeuroReport</i> , 1999 , 10, 2255-60	1.7	25
91	Genetic and epigenetic study of ALS-discordant identical twins with double mutations in SOD1 and ARHGEF28. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016 , 87, 1268-1270	5.5	25
90	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. <i>Brain</i> , 2018 , 141, 2895-2907	11.2	25
89	Mutation analysis of patients with neurodegenerative disorders using NeuroX array. <i>Neurobiology of Aging</i> , 2015 , 36, 545.e9-14	5.6	24
88	Unaffected mosaic case: RNA foci, dipeptide proteins, but upregulated C9orf72 expression. <i>Neurology</i> , 2018 , 90, e323-e331	6.5	24
87	Age-at-onset linkage analysis in Caribbean Hispanics with familial late-onset Alzheimer's disease. <i>Neurogenetics</i> , 2008 , 9, 51-60	3	24
86	Novel presenilin 1 mutations associated with early onset of dementia in a family with both early-onset and late-onset Alzheimer disease. <i>Archives of Neurology</i> , 2000 , 57, 1454-7		24
85	Late-onset vs nonmendelian early-onset Alzheimer disease: A distinction without a difference?. <i>Neurology: Genetics</i> , 2020 , 6, e512	3.8	24
84	Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. <i>Neurobiology of Aging</i> , 2016 , 42, 217.e7-217.e13	5.6	24
83	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. <i>Neurobiology of Aging</i> , 2019 , 77, 169-177	5.6	24
82	The relationship between brain atrophy and cognitive-behavioural symptoms in retired Canadian football players with multiple concussions. <i>NeuroImage: Clinical</i> , 2018 , 19, 551-558	5.3	24
81	Cerebral perfusion changes in presymptomatic genetic frontotemporal dementia: a GENFI study. <i>Brain</i> , 2019 , 142, 1108-1120	11.2	23
80	Amyloid-beta-protein isoforms in brain of subjects with PS1-linked, beta APP-linked and sporadic Alzheimer disease. <i>Molecular Brain Research</i> , 1998 , 56, 178-85		23

79	Further examination of the candidate genes in chromosome 12p13 locus for late-onset Alzheimer disease. <i>Neurogenetics</i> , 2008 , 9, 127-38	3	23
78	Homozygous and heterozygous PINK1 mutations: considerations for diagnosis and care of Parkinson's disease patients. <i>Movement Disorders</i> , 2006 , 21, 875-9	7	23
77	Clinical findings in a large family with a parkin ex3delta40 mutation. <i>Archives of Neurology</i> , 2004 , 61, 701-4		23
76	DNA Methylation Clocks and Their Predictive Capacity for Aging Phenotypes and Healthspan. <i>Neuroscience Insights</i> , 2020 , 15, 2633105520942221	3	22
75	Long-term statin therapy and CSF cholesterol levels: implications for Alzheimer's disease. <i>Dementia and Geriatric Cognitive Disorders</i> , 2009 , 27, 519-24	2.6	21
74	Early-Onset Alzheimer's Disease: What Is Missing in Research?. <i>Current Neurology and Neuroscience Reports</i> , 2021 , 21, 4	6.6	20
73	Dysregulation of chromatin remodelling complexes in amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2017 , 26, 4142-4152	5.6	19
72	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. <i>NeuroImage</i> , 2019 , 189, 645-654	7.9	18
71	Childhood onset in familial prion disease with a novel mutation in the PRNP gene. <i>Archives of Neurology</i> , 2006 , 63, 1016-21		18
70	T313M PINK1 mutation in an extended highly consanguineous Saudi family with early-onset Parkinson disease. <i>Archives of Neurology</i> , 2006 , 63, 1483-5		18
69	Clinical and genetic study of a Brazilian family with spastic paraplegia (SPG6 locus). <i>Movement Disorders</i> , 2006 , 21, 279-81	7	17
68	Inbreeding among Caribbean Hispanics from the Dominican Republic and its effects on risk of Alzheimer disease. <i>Genetics in Medicine</i> , 2015 , 17, 639-43	8.1	16
67	Intra-familial clinical heterogeneity due to FTL-DU with TDP-43 proteinopathy caused by a novel deletion in progranulin gene (PGRN). <i>Journal of Alzheimer's Disease</i> , 2010 , 22, 1123-33	4.3	16
66	Heritability and genetic variance of dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2019 , 127, 492-501	5.0	15
65	Mutation analysis of CHCHD2 in Canadian patients with familial Parkinson's disease. <i>Neurobiology of Aging</i> , 2016 , 38, 217.e7-217.e8	5.6	15
64	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. <i>Acta Neuropathologica Communications</i> , 2020 , 8, 5	7.3	15
63	Heart rate variability in leucine-rich repeat kinase 2-associated Parkinson's disease. <i>Movement Disorders</i> , 2017 , 32, 610-614	7	14
62	Marked Differences in C9orf72 Methylation Status and Isoform Expression between C9/ALS Human Embryonic and Induced Pluripotent Stem Cells. <i>Stem Cell Reports</i> , 2016 , 7, 927-940	8	14

61	The G2019S LRRK2 mutation in Brazilian patients with Parkinson's disease: phenotype in monozygotic twins. <i>Movement Disorders</i> , 2008 , 23, 290-4	7	14
60	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. <i>JAMA Network Open</i> , 2021 , 4, e2030194	10.4	14
59	Distinct biochemical signatures characterize peripherin isoform expression in both traumatic neuronal injury and motor neuron disease. <i>Journal of Neurochemistry</i> , 2010 , 114, 1177-92	6	13
58	Statins differentially affect amyloid precursor protein metabolism in presymptomatic PS1 and non-PS1 subjects. <i>Archives of Neurology</i> , 2007 , 64, 1672-3		13
57	Genetic Variations in ABCA7 Can Increase Secreted Levels of Amyloid- β 0 and Amyloid- β 2 Peptides and ABCA7 Transcription in Cell Culture Models. <i>Journal of Alzheimers Disease</i> , 2016 , 53, 875-923	4.3	13
56	White matter hyperintensities in progranulin-associated frontotemporal dementia: A longitudinal GENFI study. <i>NeuroImage: Clinical</i> , 2019 , 24, 102077	5.3	13
55	C9orf72 and ATXN2 repeat expansions coexist in a family with ataxia, dementia, and parkinsonism. <i>Movement Disorders</i> , 2017 , 32, 158-162	7	12
54	DNA methylation age acceleration is associated with ALS age of onset and survival. <i>Acta Neuropathologica</i> , 2020 , 139, 943-946	14.3	12
53	Parkinsonism due to A53E β synuclein gene mutation: Clinical, genetic, epigenetic, and biochemical features. <i>Movement Disorders</i> , 2018 , 33, 1950-1955	7	12
52	Diagnostic delay in Parkinson's disease caused by PRKN mutations. <i>Parkinsonism and Related Disorders</i> , 2019 , 63, 217-220	3.6	11
51	Mutation analysis of C9orf72 in patients with corticobasal syndrome. <i>Neurobiology of Aging</i> , 2015 , 36, 2905.e1-5	5.6	11
50	Frequent missense and insertion/deletion polymorphisms in the ovine Shadoo gene parallel species-specific variation in PrP. <i>PLoS ONE</i> , 2009 , 4, e6538	3.7	11
49	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. <i>Annals of Neurology</i> , 2020 , 88, 113-122	9.4	11
48	Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2017 , 49, 214.e13-214.e15	5.6	10
47	Absence of association between Alzheimer disease and the regulatory region polymorphism of the PS2 gene in an Italian population. <i>Neuroscience Letters</i> , 2003 , 343, 210-2	3.3	10
46	Spatiotemporal analysis for detection of pre-symptomatic shape changes in neurodegenerative diseases: Initial application to the GENFI cohort. <i>NeuroImage</i> , 2019 , 188, 282-290	7.9	10
45	A comprehensive screening of copy number variability in dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2019 , 75, 223.e1-223.e10	5.6	10
44	Comprehensive mutational analysis of LRRK2 reveals variants supporting association with autosomal dominant Parkinson's disease. <i>Journal of Human Genetics</i> , 2011 , 56, 671-5	4.3	9

43	A novel PS1 gene mutation in a large Aboriginal kindred. <i>Canadian Journal of Neurological Sciences</i> , 2010 , 37, 359-64	1	9
42	Association studies between the plasmin genes and late-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2007 , 28, 1041-3	5.6	9
41	Genomewide Association Studies of LRRK2 Modifiers of Parkinson's Disease. <i>Annals of Neurology</i> , 2021 , 90, 76-88	9.4	9
40	LRP10 in β -synucleinopathies. <i>Lancet Neurology</i> , 2018 , 17, 1033-1034	24.1	9
39	LRP10 in β -synucleinopathies. <i>Lancet Neurology</i> , 2018 , 17, 1032-1033	24.1	9
38	Parkinson's Disease, NOTCH3 Genetic Variants, and White Matter Hyperintensities. <i>Movement Disorders</i> , 2020 , 35, 2090-2095	7	8
37	Interaction of APOE4 alleles and PET tau imaging in former contact sport athletes. <i>NeuroImage: Clinical</i> , 2020 , 26, 102212	5.3	8
36	An APOE-independent cis-eSNP on chromosome 19q13.32 influences tau levels and late-onset Alzheimer's disease risk. <i>Neurobiology of Aging</i> , 2018 , 66, 178.e1-178.e8	5.6	8
35	Genetic studies of GRN and IFT74 in amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2008 , 29, 1279-88	3.8	8
34	Genetic analysis of CHCHD2 and CHCHD10 in Italian patients with Parkinson's disease. <i>Neurobiology of Aging</i> , 2017 , 53, 193.e7-193.e8	5.6	7
33	Actigraphy Detects Greater Intra-Individual Variability During Gait in Non-Manifesting LRRK2 Mutation Carriers. <i>Journal of Parkinsons Disease</i> , 2018 , 8, 131-139	5.3	7
32	Genetics and genomics of late-onset Alzheimer's disease and its endophenotypes. <i>International Journal of Alzheimers Disease</i> , 2011 , 2011, 284728	3.7	7
31	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. <i>Neuroscience Letters</i> , 2004 , 363, 99-101	3.3	7
30	Genetic markers in the diagnosis of Alzheimer's disease. <i>Journal of Alzheimers Disease</i> , 2001 , 3, 293-304	4.3	7
29	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. <i>Cortex</i> , 2020 , 133, 384-398	3.8	7
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21	Ultra-rare mutations in segregate in Caribbean Hispanic families with Alzheimer disease. <i>Neurology: Genetics</i> , 2017 , 3, e178	3.8	5
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9	Comparison of clinical and pathological phenotypes in two ethnically and geographically unrelated pedigrees segregating an equivalent presenilin 1 mutation. <i>Journal of Neuropsychiatry and Clinical Neurosciences</i> , 2000 , 12, 359-63	2.7	2
8	Combined epigenetic/genetic study identified an ALS age of onset modifier. <i>Acta Neuropathologica Communications</i> , 2021 , 9, 75	7.3	2

7	MRI-visible perivascular space volumes, sleep duration and daytime dysfunction in adults with cerebrovascular disease. <i>Sleep Medicine</i> , 2021 , 83, 83-88	4.6	2
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