Pau Pastor

List of Publications by Year in Descending Order

240 papers	12,713 citations	49 h-index	107 g-index
268 ext. papers	16,727 ext. citations	7.6 avg, IF	5.17 L-index

#	Paper	IF	Citations
240	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study.. *Movement Disorders*, **2022**,	7	2
239	Association of Essential Tremor With Novel Risk Loci: A Genome-Wide Association Study and Meta-analysis.. *JAMA Neurology*, **2022**,	17.2	3
238	Prion-like □synuclein pathology in the brain of infants with Krabbe disease.. *Brain*, **2022**,	11.2	2
237	Increased homocysteine levels correlate with cortical structural damage in Parkinson's disease.. *Journal of the Neurological Sciences*, **2022**, 434, 120148	3.2	3
236	Vitamin D Receptor and Binding Protein Gene Variants in Patients with Essential Tremor.. *Molecular Neurobiology*, **2022**, 1	6.2	
235	Smoking is associated with age at disease onset in Parkinson's disease.. *Parkinsonism and Related Disorders*, **2022**, 97, 79-83	3.6	0
234	New insights into the genetic etiology of Alzheimer's disease and related dementias.. *Nature Genetics*, **2022**,	36.3	27
233	Motor Fluctuations Development Is Associated with Non-Motor Symptoms Burden Progression in Parkinson□s Disease Patients: A 2-Year Follow-Up Study. *Diagnostics*, **2022**, 12, 1147	3.8	2
232	Constipation Predicts Cognitive Decline in Parkinson's Disease: Results from the COPPADIS Cohort at 2-Year Follow-up and Comparison with a Control Group. *Journal of Parkinson□s Disease*, **2021**,	5.3	2
231	Mendelian Randomisation Study of Smoking, Alcohol, and Coffee Drinking in Relation to Parkinson's Disease. *Journal of Parkinson□s Disease*, **2021**,	5.3	3
230	Falls Predict Acute Hospitalization in Parkinson's Disease. *Journal of Parkinson□s Disease*, **2021**,	5.3	2
229	Human-lineage-specific genomic elements are associated with neurodegenerative disease and APOE transcript usage. *Nature Communications*, **2021**, 12, 2076	17.4	1
228	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
227	Genomic Markers for Essential Tremor. *Pharmaceuticals*, **2021**, 14,	5.2	3
226	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. *Annals of Neurology*, **2021**, 90, 35-42	9.4	6
225	Staging Parkinson's Disease Combining Motor and Nonmotor Symptoms Correlates with Disability and Quality of Life. *Parkinson□s Disease*, **2021**, 2021, 8871549	2.6	6
224	Common variants in Alzheimer's disease and risk stratification by polygenic risk scores. *Nature Communications*, **2021**, 12, 3417	17.4	23

223	Identification of sixteen novel candidate genes for late onset Parkinson's disease. *Molecular Neurodegeneration*, **2021**, 16, 35	19	3
222	Predictors of Global Non-Motor Symptoms Burden Progression in Parkinson's Disease. Results from the COPPADIS Cohort at 2-Year Follow-Up. *Journal of Personalized Medicine*, **2021**, 11,	3.6	4
221	In vivo cholinergic basal forebrain degeneration and cognition in Parkinson's disease: Imaging results from the COPPADIS study. *Parkinsonism and Related Disorders*, **2021**, 88, 68-75	3.6	3
220	Mood in Parkinson's disease: From early- to late-stage disease. *International Journal of Geriatric Psychiatry*, **2021**, 36, 627-646	3.9	2
219	Genetic variation in APOE, GRN, and TP53 are phenotype modifiers in frontotemporal dementia. *Neurobiology of Aging*, **2021**, 99, 99.e15-99.e22	5.6	3
218	Added value of cerebrospinal fluid multimarker analysis in diagnosis and progression of dementia. *European Journal of Neurology*, **2021**, 28, 1142-1152	6	4
217	Serum vitamin D, vitamin D receptor and binding protein genes polymorphisms in restless legs syndrome. *Journal of Neurology*, **2021**, 268, 1461-1472	5.5	1
216	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Neuron*, **2021**, 109, 448-460.e4	13.9	20
215	Exome-wide rare variant analysis in familial essential tremor. *Parkinsonism and Related Disorders*, **2021**, 82, 109-116	3.6	5
214	Long runs of homozygosity are associated with Alzheimer's disease. *Translational Psychiatry*, **2021**, 11, 142	8.6	0
213	Common Endothelial Nitric Oxide Synthase Single Nucleotide Polymorphisms are not Related With the Risk for Restless Legs Syndrome. *Frontiers in Pharmacology*, **2021**, 12, 618989	5.6	0
212	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
211	Predictors of Loss of Functional Independence in Parkinson's Disease: Results from the COPPADIS Cohort at 2-Year Follow-Up and Comparison with a Control Group. *Diagnostics*, **2021**, 11,	3.8	2
210	Predictors of clinically significant quality of life impairment in Parkinson's disease.. *Npj Parkinsons Disease*, **2021**, 7, 118	9.7	4
209	Genomic Characterization of Host Factors Related to SARS-CoV-2 Infection in People with Dementia and Control Populations: The GR@ACE/DEGESCO Study.. *Journal of Personalized Medicine*, **2021**, 11,	3.6	2
208	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. *Nature Communications*, **2021**, 12, 7342	17.4	2
207	Multicentre, randomised, single-blind, parallel group trial to compare the effectiveness of a Holter for Parkinson's symptoms against other clinical monitoring methods: study protocol **2021**, 11, e045272		0
206	Assessing the NOTCH2NLC GGC expansion in European patients with essential tremor. *Brain*, **2020**, 143, e89	11.2	7

205	Non-motor symptom burden is strongly correlated to motor complications in patients with Parkinson's disease. *European Journal of Neurology*, **2020**, 27, 1210-1223	6	22
204	Transcriptomic differences in MSA clinical variants. *Scientific Reports*, **2020**, 10, 10310	4.9	2
203	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. *Nature Communications*, **2020**, 11, 1041	17.4	6
202	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. *Movement Disorders*, **2020**, 35, 774-780	7	27
201	The impact of freezing of gait on functional dependency in Parkinson's disease with regard to motor phenotype. *Neurological Sciences*, **2020**, 41, 2883-2892	3.5	9
200	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. *Acta Neuropathologica Communications*, **2020**, 8, 5	7.3	15
199	Disease-Specific Changes in Reelin Protein and mRNA in Neurodegenerative Diseases. *Cells*, **2020**, 9,	7.9	1
198	Role for ATXN1, ATXN2, and HTT intermediate repeats in frontotemporal dementia and Alzheimer's disease. *Neurobiology of Aging*, **2020**, 87, 139.e1-139.e7	5.6	13
197	A rare heterozygous TREM2 coding variant identified in familial clustering of dementia affects an intrinsically disordered protein region and function of TREM2. *Human Mutation*, **2020**, 41, 169-181	4.7	3
196	Genetic variability and potential effects on clinical trial outcomes: perspectives in Parkinson's disease. *Journal of Medical Genetics*, **2020**, 57, 331-338	5.8	19
195	Non-motor symptom burden in patients with Parkinson's disease with impulse control disorders and compulsive behaviours: results from the COPPADIS cohort. *Scientific Reports*, **2020**, 10, 16893	4.9	2
194	Functional genomic analyses uncover APOE-mediated regulation of brain and cerebrospinal fluid beta-amyloid levels in Parkinson disease. *Acta Neuropathologica Communications*, **2020**, 8, 196	7.3	4
193	Validity and sensitivity of instrumented postural and gait assessment using low-cost devices in Parkinson's disease. *Journal of NeuroEngineering and Rehabilitation*, **2020**, 17, 149	5.3	4
192	Mendelian randomization implies no direct causal association between leukocyte telomere length and amyotrophic lateral sclerosis. *Scientific Reports*, **2020**, 10, 12184	4.9	1
191	, age at onset, and ancestry help discriminate behavioral from language variants in FTLD cohorts. *Neurology*, **2020**, 95, e3288-e3302	6.5	5
190	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. *Movement Disorders*, **2019**, 34, 1851-1863	7	18
189	Genome-wide association analysis of dementia and its clinical endophenotypes reveal novel loci associated with Alzheimer's disease and three causality networks: The GR@ACE project. *Alzheimer's and Dementia*, **2019**, 15, 1333-1347	1.2	45
188	p.V363I mutation: A rare cause of corticobasal degeneration. *Neurology: Genetics*, **2019**, 5, e347	3.8	6

187	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. *Movement Disorders*, **2019**, 34, 460-468	7	40
186	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	14.3	50
185	Globular glial tauopathy caused by MAPT P301T mutation: clinical and neuropathological findings. *Journal of Neurology*, **2019**, 266, 2396-2405	5.5	11
184	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. *Movement Disorders*, **2019**, 34, 1333-1344	7	14
183	Cerebrospinal fluid cytokines in multiple system atrophy: A cross-sectional Catalan MSA registry study. *Parkinsonism and Related Disorders*, **2019**, 65, 3-12	3.6	12
182	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. *Npj Parkinson's Disease*, **2019**, 5, 8	9.7	47
181	COPPADIS-2015 (COhort of Patients with PArkinson's DIsease in Spain, 2015): an ongoing global Parkinson's disease project about disease progression with more than 1000 subjects included. Results from the baseline evaluation. *European Journal of Neurology*, **2019**, 26, 1399-1407	6	21
180	Genome-wide estimates of heritability and genetic correlations in essential tremor. *Parkinsonism and Related Disorders*, **2019**, 64, 262-267	3.6	8
179	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinson's disease heritability. *Npj Parkinson's Disease*, **2019**, 5, 6	9.7	51
178	Polygenic risk and hazard scores for Alzheimer's disease prediction. *Annals of Clinical and Translational Neurology*, **2019**, 6, 456-465	5.3	36
177	Heritability and genetic variance of dementia with Lewy bodies. *Neurobiology of Disease*, **2019**, 127, 492-501	7.3	15
176	Genetic variation across RNA metabolism and cell death gene networks is implicated in the semantic variant of primary progressive aphasia. *Scientific Reports*, **2019**, 9, 10854	4.9	5
175	Non-motor symptoms burden, mood, and gait problems are the most significant factors contributing to a poor quality of life in non-demented Parkinson's disease patients: Results from the COPPADIS Study Cohort. *Parkinsonism and Related Disorders*, **2019**, 66, 151-157	3.6	39
174	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. *Lancet Neurology, The*, **2019**, 18, 1091-1102	24.1	562
173	High ultrasensitive serum C-reactive protein may be related to freezing of gait in Parkinson's disease patients. *Journal of Neural Transmission*, **2019**, 126, 1599-1608	4.3	9
172	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
171	Plasma levels of soluble TREM2 and neurofilament light chain in TREM2 rare variant carriers. *Alzheimer's Research and Therapy*, **2019**, 11, 94	9	15
170	A comprehensive screening of copy number variability in dementia with Lewy bodies. *Neurobiology of Aging*, **2019**, 75, 223.e1-223.e10	5.6	10

169	Identification of evolutionarily conserved gene networks mediating neurodegenerative dementia. *Nature Medicine*, **2019**, 25, 152-164	50.5	55
168	Association between the missense alcohol dehydrogenase rs1229984T variant with the risk for Parkinson's disease in women. *Journal of Neurology*, **2019**, 266, 346-352	5.5	9
167	Dynamic Atlas-Based Segmentation and Quantification of Neuromelanin-Rich Brainstem Structures in Parkinson Disease. *IEEE Transactions on Medical Imaging*, **2019**, 38, 813-823	11.7	19
166	CXCR4 involvement in neurodegenerative diseases. *Translational Psychiatry*, **2018**, 8, 73	8.6	36
165	Target-enriched sequencing of chromosome 17q21.31 in sporadic tauopathies reveals no candidate variants. *Neurobiology of Aging*, **2018**, 66, 177.e7-177.e10	5.6	1
164	Gamma-aminobutyric acid (GABA) receptors genes polymorphisms and risk for restless legs syndrome. *Pharmacogenomics Journal*, **2018**, 18, 565-577	3.5	10
163	Rare nonsynonymous variants in SORT1 are associated with increased risk for frontotemporal dementia. *Neurobiology of Aging*, **2018**, 66, 181.e3-181.e10	5.6	12
162	Pleiotropic Effects of Variants in Dementia Genes in Parkinson Disease. *Frontiers in Neuroscience*, **2018**, 12, 230	5.1	11
161	Pooled-DNA target sequencing of Parkinson genes reveals novel phenotypic associations in Spanish population. *Neurobiology of Aging*, **2018**, 70, 325.e1-325.e5	5.6	5
160	Systematic Screening of Ubiquitin/p62 Aggregates in Cerebellar Cortex Expands the Neuropathological Phenotype of the C9orf72 Expansion Mutation. *Journal of Neuropathology and Experimental Neurology*, **2018**, 77, 703-709	3.1	8
159	No supportive evidence for TIA1 gene mutations in a European cohort of ALS-FTD spectrum patients. *Neurobiology of Aging*, **2018**, 69, 293.e9-293.e11	5.6	11
158	Immune-related genetic enrichment in frontotemporal dementia: An analysis of genome-wide association studies. *PLoS Medicine*, **2018**, 15, e1002487	11.6	77
157	Clinic-Based Validation of Cerebrospinal Fluid Biomarkers with Florbetapir PET for Diagnosis of Dementia. *Journal of Alzheimer's Disease*, **2018**, 61, 135-143	4.3	8
156	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
155	Cerebrospinal fluid levels of coenzyme Q10 are reduced in multiple system atrophy. *Parkinsonism and Related Disorders*, **2018**, 46, 16-23	3.6	15
154	Common and rare TBK1 variants in early-onset Alzheimer disease in a European cohort. *Neurobiology of Aging*, **2018**, 62, 245.e1-245.e7	5.6	12
153	LRP10 in α-synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032	24.1	14
152	LRP10 in α-synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032-1033	24.1	9

151	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. *Brain*, **2018**, 141, 2895-2907	11.2	25
150	Genetically elevated high-density lipoprotein cholesterol through the cholesteryl ester transfer protein gene does not associate with risk of Alzheimer's disease. *Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring*, **2018**, 10, 595-598	5.2	
149	Heterozygous mutation causes familial ataxia with cognitive affective syndrome (SCA48). *Neurology*, **2018**, 91, e1988-e1998	6.5	59
148	Genetic architecture of sporadic frontotemporal dementia and overlap with Alzheimer's and Parkinson's diseases. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, 152-164	5.5	76
147	association with REM sleep behavior disorder. *Neurology: Genetics*, **2017**, 3, e131	3.8	10
146	Deleterious ABCA7 mutations and transcript rescue mechanisms in early onset Alzheimer's disease. *Acta Neuropathologica*, **2017**, 134, 475-487	14.3	34
145	Phosphorylated neurofilament heavy chain: A biomarker of survival for C9ORF72-associated amyotrophic lateral sclerosis. *Annals of Neurology*, **2017**, 82, 139-146	9.4	58
144	Delta-amino-levulinic acid dehydratase gene and essential tremor. *European Journal of Clinical Investigation*, **2017**, 47, 348-356	4.6	4
143	TBK1 Mutation Spectrum in an Extended European Patient Cohort with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Human Mutation*, **2017**, 38, 297-309	4.7	66
142	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
141	Lack of evidence for a role of genetic variation in TMEM230 in the risk for Parkinson's disease in the Caucasian population. *Neurobiology of Aging*, **2017**, 50, 167.e11-167.e13	5.6	22
140	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
139	Thr105Ile (rs11558538) polymorphism in the histamine-1-methyl-transferase (HNMT) gene and risk for restless legs syndrome. *Journal of Neural Transmission*, **2017**, 124, 285-291	4.3	9
138	[O1□1□3]: CEREBROSPINAL FLUID ENDOPHENOTYPES PROVIDE INSIGHT INTO BIOLOGY UNDERLYING ALZHEIMER'S DISEASE **2017**, 13, P218-P219		
137	Association Between the rs1229984 Polymorphism in the Alcohol Dehydrogenase 1B Gene and Risk for Restless Legs Syndrome. *Sleep*, **2017**, 40,	1.1	9
136	Discovering the 3' UTR-mediated regulation of alpha-synuclein. *Nucleic Acids Research*, **2017**, 45, 12888-12903	10.1	23
135	A novel Alzheimer disease locus located near the gene encoding tau protein. *Molecular Psychiatry*, **2016**, 21, 108-17	15.1	175
134	Heme Oxygenase-1 and 2 Common Genetic Variants and Risk for Multiple Sclerosis. *Scientific Reports*, **2016**, 6, 20830	4.9	17

133	Genome-wide association study in essential tremor identifies three new loci. *Brain*, **2016**, 139, 3163-3169	11.2	57
132	Genetics of Essential Tremor **2016**, 1-14		
131	Resequencing analysis of five Mendelian genes and the top genes from genome-wide association studies in Parkinson's Disease. *Molecular Neurodegeneration*, **2016**, 11, 29	19	46
130	COPPADIS-2015 (COhort of Patients with PArkinson's DIsease in Spain, 2015), a global--clinical evaluations, serum biomarkers, genetic studies and neuroimaging--prospective, multicenter, non-interventional, long-term study on Parkinson's disease progression. *BMC Neurology*, **2016**, 16, 26	3.1	46
129	Copy number variation analysis of the 17q21.31 region and its role in neurodegenerative diseases. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2016**, 171B, 175-80	3.5	11
128	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
127	Assessing the role of TUBA4A gene in frontotemporal degeneration. *Neurobiology of Aging*, **2016**, 38, 215.e13-215.e14	5.6	7
126	MAPT H1 Haplotype is Associated with Late-Onset Alzheimer's Disease Risk in APOEe4 Noncarriers: Results from the Dementia Genetics Spanish Consortium. *Journal of Alzheimer's Disease*, **2016**, 49, 343-52	4.3	26
125	Neuroimaging Correlates of Frontotemporal Dementia Associated with SQSTM1 Mutations. *Journal of Alzheimer's Disease*, **2016**, 53, 303-13	4.3	8
124	A comprehensive study of the genetic impact of rare variants in SORL1 in European early-onset Alzheimer's disease. *Acta Neuropathologica*, **2016**, 132, 213-224	14.3	62
123	A family study of DRD3 rs6280, SLC1A2 rs3794087 and MAPT rs1052553 variants in essential tremor. *Neurological Research*, **2016**, 38, 880-7	2.7	7
122	Shared genetic contribution to Ischaemic Stroke and Alzheimer's Disease. *Annals of Neurology*, **2016**, 79, 739-747	9.4	42
121	Aberrant mitochondria in a Bethlem myopathy patient with a homozygous amino acid substitution that destabilizes the collagen VI α2(VI) chain. *Journal of Biological Chemistry*, **2015**, 290, 4272-81	5.4	20
120	Missense mutations in TENM4, a regulator of axon guidance and central myelination, cause essential tremor. *Human Molecular Genetics*, **2015**, 24, 5677-86	5.6	83
119	Analysis of the CHCHD10 gene in patients with frontotemporal dementia and amyotrophic lateral sclerosis from Spain. *Brain*, **2015**, 138, e400	11.2	47
118	Genetic variability in SQSTM1 and risk of early-onset Alzheimer dementia: a European early-onset dementia consortium study. *Neurobiology of Aging*, **2015**, 36, 2005.e15-22	5.6	29
117	TREM2 R47H variant and risk of essential tremor: a cross-sectional international multicenter study. *Parkinsonism and Related Disorders*, **2015**, 21, 306-9	3.6	26
116	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146

115	Clinical and neuroimaging characteristics of 14 patients with prionopathy: a descriptive study. *Neurologí*, **2015**, 30, 144-52	1.4	7
114	No evidence of association between common European mitochondrial DNA variants in Alzheimer, Parkinson, and migraine in the Spanish population. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2015**, 168B, 54-65	3.5	33
113	O3-13-06: Targeted re-sequencing of sorl1 in early-onset Alzheimer's dementia: The european early onset dementia consortium **2015**, 11, P253-P253		
112	Rare Variants in PLD3 Do Not Affect Risk for Early-Onset Alzheimer Disease in a European Consortium Cohort. *Human Mutation*, **2015**, 36, 1226-35	4.7	20
111	P2-049: Functional characterization of a novel TREM2 coding variant linked to familial Alzheimer's disease **2015**, 11, P500-P500		2
110	Association Between Vitamin D Receptor rs731236 (Taq1) Polymorphism and Risk for Restless Legs Syndrome in the Spanish Caucasian Population. *Medicine (United States)*, **2015**, 94, e2125	1.8	18
109	Automated neuromelanin imaging as a diagnostic biomarker for Parkinson's disease. *Movement Disorders*, **2015**, 30, 945-52	7	93
108	Heme Oxygenase 1 and 2 Common Genetic Variants and Risk for Essential Tremor. *Medicine (United States)*, **2015**, 94, e968	1.8	17
107	Heme Oxygenase-1 and 2 Common Genetic Variants and Risk for Restless Legs Syndrome. *Medicine (United States)*, **2015**, 94, e1448	1.8	28
106	Atlas-based segmentation of brainstem regions in neuromelanin-sensitive magnetic resonance images **2015**,		1
105	Rare variants in Amyloid precursor protein (APP) and Parkinson's disease. *European Journal of Human Genetics*, **2015**, 23, 1328-33	5.3	29
104	Characterization of the repeat expansion size in C9orf72 in amyotrophic lateral sclerosis and frontotemporal dementia. *Human Molecular Genetics*, **2014**, 23, 749-54	5.6	84
103	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. *Nature*, **2014** , 505, 550-554	50.4	345
102	Protective effect of LRRK2 p.R1398H on risk of Parkinson's disease is independent of MAPT and SNCA variants. *Neurobiology of Aging*, **2014**, 35, 266.e5-14	5.6	26
101	Investigation of the role of rare TREM2 variants in frontotemporal dementia subtypes. *Neurobiology of Aging*, **2014**, 35, 2657.e13-2657.e19	5.6	31
100	Rare mutations in SQSTM1 modify susceptibility to frontotemporal lobar degeneration. *Acta Neuropathologica*, **2014**, 128, 397-410	14.3	83
99	Frontotemporal dementia and its subtypes: a genome-wide association study. *Lancet Neurology, The*, **2014**, 13, 686-99	24.1	207
98	Analysis of nuclear export sequence regions of FUS-Related RNA-binding proteins in essential tremor. *PLoS ONE*, **2014**, 9, e111989	3.7	8

97	An association study between Heme oxygenase-1 genetic variants and Parkinson's disease. *Frontiers in Cellular Neuroscience*, **2014**, 8, 298	6.1	29
96	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
95	Global investigation and meta-analysis of the C9orf72 (G4C2)n repeat in Parkinson disease. *Neurology*, **2014**, 83, 1906-13	6.5	49
94	Frontobasal gray matter loss is associated with the TREM2 p.R47H variant. *Neurobiology of Aging*, **2014**, 35, 2681-2690	5.6	31
93	Assessing the role of the TREM2 p.R47H variant as a risk factor for Alzheimer's disease and frontotemporal dementia. *Neurobiology of Aging*, **2014**, 35, 444.e1-4	5.6	81
92	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
91	LINGO1 rs9652490 and rs11856808 polymorphisms are not associated with risk for multiple sclerosis. *BMC Neurology*, **2013**, 13, 34	3.1	7
90	The unfolded protein response is activated in disease-affected brain regions in progressive supranuclear palsy and Alzheimer's disease. *Acta Neuropathologica Communications*, **2013**, 1, 31	7.3	104
89	Mutational screening of PARKIN identified a 3' UTR variant (rs62637702) associated with Parkinson's disease. *Journal of Molecular Neuroscience*, **2013**, 50, 264-9	3.3	8
88	Automated analysis of FDG PET as a tool for single-subject probabilistic prediction and detection of Alzheimer's disease dementia. *European Journal of Nuclear Medicine and Molecular Imaging*, **2013**, 40, 1394-405	8.8	38
87	Genetic risk score predicting accelerated progression from mild cognitive impairment to Alzheimer's disease. *Journal of Neural Transmission*, **2013**, 120, 807-12	4.3	58
86	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
85	Orthostatic myoclonus: an underrecognized cause of unsteadiness?. *Parkinsonism and Related Disorders*, **2013**, 19, 1013-7	3.6	16
84	TREM2 is associated with the risk of Alzheimer's disease in Spanish population. *Neurobiology of Aging*, **2013**, 34, 1711.e15-7	5.6	121
83	TREM2 and neurodegenerative disease. *New England Journal of Medicine*, **2013**, 369, 1568-9	59.2	89
82	Analysis of the C9orf72 gene in patients with amyotrophic lateral sclerosis in Spain and different populations worldwide. *Human Mutation*, **2013**, 34, 79-82	4.7	71
81	Fused in Sarcoma (FUS) gene mutations are not a frequent cause of essential tremor in Europeans. *Neurobiology of Aging*, **2013**, 34, 2441.e9-2441.e11	5.6	21
80	Update on genetics of essential tremor. *Acta Neurologica Scandinavica*, **2013**, 128, 359-71	3.8	38

79	Selective brain gray matter atrophy associated with APOE ε4 and MAPT H1 in subjects with mild cognitive impairment. *Journal of Alzheimer's Disease*, **2013**, 33, 1009-19	4.3	17
78	No association of the SLC1A2 rs3794087 allele with risk for essential tremor in the Spanish population. *Pharmacogenetics and Genomics*, **2013**, 23, 587-90	1.9	18
77	Comment: double mutants of frontotemporal dementia genes--Simple co-occurrence?. *Neurology*, **2013**, 81, 1338	6.5	3
76	C9ORF72 repeat expansion in Australian and Spanish frontotemporal dementia patients. *PLoS ONE*, **2013**, 8, e56899	3.7	51
75	Dopaminergic neuronal imaging in genetic Parkinson's disease: insights into pathogenesis. *PLoS ONE*, **2013**, 8, e69190	3.7	46
74	Rare variants in calcium homeostasis modulator 1 (CALHM1) found in early onset Alzheimer's disease patients alter calcium homeostasis. *PLoS ONE*, **2013**, 8, e74203	3.7	22
73	LRRK2 haplotype-sharing analysis in Parkinson's disease reveals a novel p.S1761R mutation. *Movement Disorders*, **2012**, 27, 146-51	7	15
72	Common variation in the LRRK2 gene is a risk factor for Parkinson's disease. *Movement Disorders*, **2012**, 27, 1822-5	7	12
71	Nigrostriatal dopaminergic function in subjects with isolated action tremor. *Parkinsonism and Related Disorders*, **2012**, 18, 49-53	3.6	21
70	LINGO1 rs9652490 and rs11856808 are not associated with the risk of Parkinson's disease: results of a meta-analysis. *Parkinsonism and Related Disorders*, **2012**, 18, 657-9	3.6	7
69	LINGO1 and risk for essential tremor: results of a meta-analysis of rs9652490 and rs11856808. *Journal of the Neurological Sciences*, **2012**, 317, 52-7	3.2	35
68	Age at onset in LRRK2-associated PD is modified by SNCA variants. *Journal of Molecular Neuroscience*, **2012**, 48, 245-7	3.3	31
67	Pooled-DNA sequencing identifies novel causative variants in PSEN1, GRN and MAPT in a clinical early-onset and familial Alzheimer's disease Ibero-American cohort. *Alzheimer's Research and Therapy*, **2012**, 4, 34	9	78
66	H1-MAPT and the risk for familial essential tremor. *PLoS ONE*, **2012**, 7, e41581	3.7	15
65	A search for SNCA 3' UTR variants identified SNP rs356165 as a determinant of disease risk and onset age in Parkinson's disease. *Journal of Molecular Neuroscience*, **2012**, 47, 425-30	3.3	36
64	Usefulness of positron emission tomography with fludeoxyglucose f 18 and with carbon 11-tagged methionine in the diagnosis of hippocampal lesions. *Archives of Neurology*, **2012**, 69, 1652-3		
63	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. *Nature Genetics*, **2011**, 43, 699-705	36.3	386
62	Different MAPT haplotypes are associated with Parkinson's disease and progressive supranuclear palsy. *Neurobiology of Aging*, **2011**, 32, 547.e11-6	5.6	25

61	Genetic variation in APOE cluster region and Alzheimer's disease risk. *Neurobiology of Aging*, **2011**, 32, 2107.e7-17	5.6	31
60	Isolated dysphagia due to paraneoplastic myasthenic syndrome with anti-P/Q-type voltage-gated calcium-channel and anti-acetylcholine receptor antibodies. *Neuromuscular Disorders*, **2011**, 21, 126-8	2.9	6
59	Gamma-aminobutyric acid GABRA4, GABRE, and GABRQ receptor polymorphisms and risk for essential tremor. *Pharmacogenetics and Genomics*, **2011**, 21, 436-9	1.9	25
58	A polymorphism located at an ATG transcription start site of the heme oxygenase-2 gene is associated with classical Parkinson's disease. *Pharmacogenetics and Genomics*, **2011**, 21, 565-71	1.9	12
57	Lack of interaction of SNCA and MAPT genotypes in Parkinson's disease. *European Journal of Neurology*, **2011**, 18, e32	6	12
56	Lack of association of LINGO1 rs9652490 and rs11856808 SNPs with familial essential tremor. *European Journal of Neurology*, **2011**, 18, 1085-9	6	27
55	Gamma-aminobutyric acid (GABA) receptor rho (GABRR) polymorphisms and risk for essential tremor. *Journal of Neurology*, **2011**, 258, 203-11	5.5	25
54	LINGO1 gene analysis in Parkinson's disease phenotypes. *Movement Disorders*, **2011**, 26, 722-7	7	15
53	Replication of MAPT and SNCA, but not PARK16-18, as susceptibility genes for Parkinson's disease. *Movement Disorders*, **2011**, 26, 819-23	7	55
52	The effect of MAPT H1 and APOE ε4 on transition from mild cognitive impairment to dementia. *Journal of Alzheimer's Disease*, **2010**, 22, 1065-71	4.3	21
51	PINK1-linked parkinsonism is associated with Lewy body pathology. *Brain*, **2010**, 133, 1128-42	11.2	188
50	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
49	Spastic Paraparesis **2010**, 132-139		1
48	Analysis of the Micro-RNA-133 and PITX3 genes in Parkinson's disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1234-9	3.5	25
47	Analysis of the GIGYF2 gene in familial and sporadic Parkinson disease in the Spanish population. *European Journal of Neurology*, **2010**, 17, 321-5	6	7
46	Cortical atrophy and language network reorganization associated with a novel progranulin mutation. *Cerebral Cortex*, **2009**, 19, 1751-60	5.1	42
45	Familial neurodegeneration in progressive supranuclear palsy: more frequent than expected?. *Neurology*, **2009**, 73, 86-7	6.5	3
44	5'-Upstream variants of CRHR1 and MAPT genes associated with age at onset in progressive supranuclear palsy and cortical basal degeneration. *Neurobiology of Disease*, **2009**, 33, 164-70	7.5	24

43	Early detection of patients in the pre demented stage of Alzheimer's disease: the Pre-Al Study. *Journal of Nutrition, Health and Aging*, **2009**, 13, 21-6	5.2	16
42	Forceps minor region signal abnormality "ears of the lynx": an early MRI finding in spastic paraparesis with thin corpus callosum and mutations in the spatacsin gene (SPG11) on chromosome 15. *Journal of Neuroimaging*, **2009**, 19, 52-60	2.8	31
41	Ubiquitin associated protein 1 is a risk factor for frontotemporal lobar degeneration. *Neurobiology of Aging*, **2009**, 30, 656-65	5.6	29
40	SPG11 compound mutations in spastic paraparesis with thin corpus callosum. *Neurology*, **2008**, 71, 332-6	6.5	27
39	Memory decline evolves independently of disease activity in MS. *Multiple Sclerosis Journal*, **2008**, 14, 947-53	5	45
38	HDDD2 is a familial frontotemporal lobar degeneration with ubiquitin-positive, tau-negative inclusions caused by a missense mutation in the signal peptide of progranulin. *Annals of Neurology*, **2006**, 60, 314-22	9.4	174
37	Amygdalar and hippocampal MRI volumetric reductions in Parkinson's disease with dementia. *Movement Disorders*, **2005**, 20, 540-4	7	109
36	Structural brain changes in Parkinson disease with dementia: a voxel-based morphometry study. *Archives of Neurology*, **2005**, 62, 281-5		238
35	A novel mutation (K317M) in the MAPT gene causes FTDP and motor neuron disease. *Neurology*, **2005**, 64, 1578-85	6.5	88
34	Identification of Genes that Modify the Age of Onset in a Large Familial Alzheimer's Disease Kindred. *Research and Perspectives in Alzheimer's Disease*, **2005**, 61-71		
33	Molecular genetics of Alzheimer's disease. *Current Psychiatry Reports*, **2004**, 6, 125-33	9.1	58
32	Tau gene delN296 mutation, Parkinson's disease, and atypical supranuclear palsy. *Annals of Neurology*, **2004**, 55, 448-9	9.4	9
31	Novel haplotypes in 17q21 are associated with progressive supranuclear palsy. *Annals of Neurology*, **2004**, 56, 249-58	9.4	59
30	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
29	A novel mutation in the PSEN2 gene (T430M) associated with variable expression in a family with early-onset Alzheimer disease. *Archives of Neurology*, **2003**, 60, 1149-51		37
28	Apolipoprotein Eepsilon4 modifies Alzheimer's disease onset in an E280A PS1 kindred. *Annals of Neurology*, **2003**, 54, 163-9	9.4	133
27	Tau phosphorylation and kinase activation in familial tauopathy linked to deln296 mutation. *Neuropathology and Applied Neurobiology*, **2003**, 29, 23-34	5.2	38
26	Further extension of the H1 haplotype associated with progressive supranuclear palsy. *Movement Disorders*, **2002**, 17, 550-6	7	47

25	Analysis of the exon 1 polymorphism in the Tau gene in transmissible spongiform encephalopathies. *Journal of Neurology*, **2002**, 249, 938-9	5.5	4
24	Dementia in Parkinson disease: a proton magnetic resonance spectroscopy study. *Archives of Neurology*, **2002**, 59, 1415-20		48
23	Relative high frequency of the c.255delA parkin gene mutation in Spanish patients with autosomal recessive parkinsonism. *Journal of Neurology, Neurosurgery and Psychiatry*, **2002**, 73, 582-4	5.5	21
22	Progressive supranuclear palsy: clinical and genetic aspects. *Current Opinion in Neurology*, **2002**, 15, 429-37	7.1	17
21	Analysis of the coding and the 5' flanking regions of the alpha-synuclein gene in patients with Parkinson's disease. *Movement Disorders*, **2001**, 16, 1115-9	7	15
20	Familial atypical progressive supranuclear palsy associated with homozigosity for the delN296 mutation in the tau gene. *Annals of Neurology*, **2001**, 49, 263-7	9.4	165
19	Examination of motor output pathways in patients with corticobasal ganglionic degeneration using transcranial magnetic stimulation. *Brain*, **2001**, 124, 1131-7	11.2	30
18	A novel presenilin 2 gene mutation (D439A) in a patient with early-onset Alzheimer's disease. *Neurology*, **2001**, 57, 1926-8	6.5	27
17	Enhanced gain of blink reflex responses to ipsilateral supraorbital nerve afferent inputs in patients with facial nerve palsy. *Clinical Neurophysiology*, **2001**, 112, 153-6	4.3	37
16	Significant association between the tau gene A0/A0 genotype and Parkinson's disease. *Annals of Neurology*, **2000**, 47, 242-245	9.4	111
15	A new mutation in the parkin gene in a patient with atypical autosomal recessive juvenile parkinsonism. *Neuroscience Letters*, **2000**, 289, 66-8	3.3	26
14	Significant association between the tau gene A0/A0 genotype and Parkinson's disease. *Annals of Neurology*, **2000**, 47, 242-5	9.4	29
13	Hyperkalemic periodic paralysis associated with multiple sleep onset REM periods. *Sleep*, **1999**, 22, 1123-4	4.1	3
12	Dopamine receptor D2 intronic polymorphism in patients with Parkinson's disease. *Neuroscience Letters*, **1999**, 273, 151-4	3.3	15
11	Identification of a novel polymorphism in the promoter region of the tau gene highly associated to progressive supranuclear palsy in humans. *Neuroscience Letters*, **1999**, 275, 183-6	3.3	52
10	Alpha1-antichymotrypsin gene polymorphism and susceptibility to Parkinson's disease. *Neurology*, **1999**, 52, 297-301	6.5	13
9	Recruitment curve of the soleus H reflex in patients with neurogenic claudication. *Muscle and Nerve* , **1998**, 21, 985-90	3.4	18
8	Identification of Spanish familial Parkinson's disease and screening for the Ala53Thr mutation of the alpha-synuclein gene in early onset patients. *Neuroscience Letters*, **1997**, 235, 57-60	3.3	61

7	Pleural and peritoneal leishmaniasis in an AIDS patient. *European Journal of Clinical Microbiology and Infectious Diseases*, **1997**, 16, 246-8	5.3	17
6	Reversible oral-facial dyskinesia in a patient receiving ciprofloxacin hydrochloride. *Journal of Neurology*, **1996**, 243, 616-7	5.5	23
5	The genetic architecture of Parkinson disease in Spain: characterizing population-specific risk, differential haplotype structures, and providing etiologic insight		1
4	Common variants in Alzheimer disease: Novel association of six genetic variants with AD and risk stratification by polygenic risk scores		9
3	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer Disease		1
2	New insights on the genetic etiology of Alzheimer and related dementia		25
1	Genome-wide association analysis of dementia and its clinical endophenotypes reveal novel loci associated with Alzheimer disease and three causality networks of AD: the GR@ACE project		3