Jose T Bras

List of Publications by Year in Descending Order

154 papers	13,367 citations	48 h-index	115 g-index
176 ext. papers	17,684 ext. citations	10 avg, IF	5.52 L-index

#	Paper	IF	Citations
154	Cognitive composites for genetic frontotemporal dementia: GENFI-Cog.. *Alzheimer's Research and Therapy*, **2022**, 14, 10	9	0
153	Prion-like α-synuclein pathology in the brain of infants with Krabbe disease.. *Brain*, **2022**,	11.2	2
152	Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort.. *Cortex*, **2022**, 150, 12-28	3.8	
151	Challenge accepted: uncovering the role of rare genetic variants in Alzheimer's disease.. *Molecular Neurodegeneration*, **2022**, 17, 3	19	1
150	Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank.. *Scientific Reports*, **2022**, 12, 8404	4.9	0
149	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. *Alzheimer's and Dementia*, **2021**,	1.2	2
148	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum NfL and pNfH: A Longitudinal Multicentre Study. *Annals of Neurology*, **2021**,	9.4	2
147	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. *Molecular Neurodegeneration*, **2021**, 16, 79	19	0
146	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. *Brain*, **2021**,	11.2	3
145	How understudied populations have contributed to our understanding of Alzheimer's disease genetics. *Brain*, **2021**, 144, 1067-1081	11.2	2
144	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. *JAMA Neurology*, **2021**, 78, 464-472	17.2	17
143	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. *Annals of Neurology*, **2021**, 90, 35-42	9.4	6
142	Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. *Neuromuscular Disorders*, **2021**, 31, 891-895	2.9	
141	Identification of sixteen novel candidate genes for late onset Parkinson's disease. *Molecular Neurodegeneration*, **2021**, 16, 35	19	3
140	Genetics of synucleins in neurodegenerative diseases. *Acta Neuropathologica*, **2021**, 141, 471-490	14.3	6
139	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. *European Journal of Human Genetics*, **2021**, 29, 663-671	5.3	0
138	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. *Alzheimer's and Dementia*, **2021**, 17, 500-514	1.2	8

137	Genetic variants in glutamate-, A and tau-related pathways determine polygenic risk for Alzheimer's disease. *Neurobiology of Aging*, **2021**, 101, 299.e13-299.e21	5.6	3
136	A Non-APOE Polygenic Risk Score for Alzheimer's Disease Is Associated With Cerebrospinal Fluid Neurofilament Light in a Representative Sample of Cognitively Unimpaired 70-Year Olds. *Journals of Gerontology - Series A Biological Sciences and Medical Sciences*, **2021**, 76, 983-990	6.4	4
135	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. *JAMA Network Open*, **2021**, 4, e2030194	10.4	14
134	A comprehensive analysis of copy number variation in a Turkish dementia cohort. *Human Genomics*, **2021**, 15, 48	6.8	
133	Disease-related cortical thinning in presymptomatic granulin mutation carriers. *NeuroImage: Clinical* , **2021**, 29, 102540	5.3	2
132	Polygenic risk scores for Alzheimer's disease are related to dementia risk in APOE e4 negatives. *Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring*, **2021**, 13, e12142	5.2	5
131	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. *Nature Communications*, **2021**, 12, 7342	17.4	2
130	Clinical, ocular motor, and imaging profile of Niemann-Pick type C heterozygosity. *Neurology*, **2020**, 94, e1702-e1715	6.5	12
129	KCNN2 mutation in autosomal-dominant tremulous myoclonus-dystonia. *European Journal of Neurology*, **2020**, 27, 1471-1477	6	10
128	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
127	Alzheimer Disease Genetics: Review of Novel Loci Associated with Disease. *Current Genetic Medicine Reports*, **2020**, 8, 1-16	2.2	12
126	Novel Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1
125	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. *Acta Neuropathologica Communications*, **2020**, 8, 5	7.3	15
124	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. *Cortex*, **2020**, 133, 384-398	3.8	7
123	Genetic architecture of common non-Alzheimer's disease dementias. *Neurobiology of Disease*, **2020**, 142, 104946	7.5	16
122	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
121	Two pathologically confirmed cases of novel mutations in the MAPT gene causing frontotemporal dementia. *Neurobiology of Aging*, **2020**, 87, 141.e15-141.e20	5.6	2
120	CYLD variants in frontotemporal dementia associated with severe memory impairment in a Portuguese cohort. *Brain*, **2020**, 143, e67	11.2	5

119	Patients with progranulin mutations overlap with the progressive dysexecutive syndrome: towards the definition of a frontoparietal dementia phenotype. *Brain Communications*, **2020**, 2, fcaa126	4.5	3
118	Psychiatric Manifestations of Mutations. *Movement Disorders Clinical Practice*, **2020**, 7, 838-841	2.2	4
117	A deletion of IDUA exon 10 in a family of Golden Retriever dogs with an attenuated form of mucopolysaccharidosis type I. *Journal of Veterinary Internal Medicine*, **2020**, 34, 1813-1824	3.1	1
116	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. *Brain Communications*, **2020**, 2,	4.5	6
115	A rare loss-of-function variant of ADAM17 is associated with late-onset familial Alzheimer disease. *Molecular Psychiatry*, **2020**, 25, 629-639	15.1	27
114	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
113	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
112	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. *Movement Disorders*, **2019**, 34, 460-468	7	40
111	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. *NeuroImage*, **2019**, 189, 645-654	7.9	18
110	Alzheimer's disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. *Translational Psychiatry*, **2019**, 9, 154	8.6	31
109	Microdeletion in a FAAH pseudogene identified in a patient with high anandamide concentrations and pain insensitivity. *British Journal of Anaesthesia*, **2019**, 123, e249-e253	5.4	51
108	Heritability and genetic variance of dementia with Lewy bodies. *Neurobiology of Disease*, **2019**, 127, 492-501	5.1	15
107	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
106	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. *Lancet Neurology, The*, **2019**, 18, 1103-1111	24.1	68
105	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
104	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. *Acta Neuropathologica Communications*, **2019**, 7, 188	7.3	6
103	Peripheral GRN mRNA and Serum Progranulin Levels as a Potential Indicator for Both the Presence of Splice Site Mutations and Individuals at Risk for Frontotemporal Dementia. *Journal of Alzheimer's Disease*, **2019**, 67, 159-167	4.3	7
102	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. *Neurobiology of Aging*, **2019**, 77, 169-177	5.6	24

101	A comprehensive screening of copy number variability in dementia with Lewy bodies. *Neurobiology of Aging*, **2019**, 75, 223.e1-223.e10	5.6	10
100	Genetics of dementia in a Finnish cohort. *European Journal of Human Genetics*, **2018**, 26, 827-837	5.3	5
99	Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. *Neurology: Genetics*, **2018**, 4, e215	3.8	8
98	A novel human pain insensitivity disorder caused by a point mutation in ZFHX2. *Brain*, **2018**, 141, 365-376	11.2	23
97	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. *Neurobiology of Aging*, **2018**, 66, 179.e17-179.e29	5.6	23
96	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. *Neurobiology of Aging*, **2018**, 62, 244.e1-244.e8	5.6	25
95	Genotyping of the Alzheimer's Disease Genome-Wide Association Study Index Single Nucleotide Polymorphisms in the Brains for Dementia Research Cohort. *Journal of Alzheimer's Disease*, **2018**, 64, 355-362	4.3	6
94	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. *JAMA Neurology*, **2018**, 75, 1416-1422	17.2	50
93	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
92	An Aged Canid with Behavioral Deficits Exhibits Blood and Cerebrospinal Fluid Amyloid Beta Oligomers. *Frontiers in Aging Neuroscience*, **2018**, 10, 7	5.3	7
91	The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. *Lancet Neurology, The*, **2018**, 17, 721-730	24.1	92
90	The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. *Current Neurology and Neuroscience Reports*, **2018**, 18, 67	6.6	41
89	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
88	LRP10 in □synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032	24.1	14
87	Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddon's Syndrome of Unknown Cause. *Cerebrovascular Diseases*, **2018**, 46, 257-264	3.2	10
86	LRP10 in □synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032-1033	24.1	9
85	splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. *Neurology: Genetics*, **2018**, 4, e273	3.8	4
84	Action Myoclonus and Seizure in Kufor-Rakeb Syndrome. *Movement Disorders Clinical Practice*, **2018**, 5, 195-199	2.2	11

83	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. *Genetics in Medicine*, **2017**, 19, 45-52	8.1	68
82	CLN8 disease caused by large genomic deletions. *Molecular Genetics & Genomic Medicine*, **2017**, 5, 85-91	2.3	4
81	Mouse models of kufor-rakeb disease link Parkinson's disease closer to neuronal ceroid lipofuscinosis, suggesting lysosomal dysfunction as shared mechanism. *Movement Disorders*, **2017**, 32, 209	7	
80	Ataxia with oculomotor apraxia is associated with the DNA damage repair pathway. *Movement Disorders*, **2017**, 32, 720	7	1
79	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
78	Diagnosis and management of dementia with Lewy bodies: Fourth consensus report of the DLB Consortium. *Neurology*, **2017**, 89, 88-100	6.5	1691
77	Multi-infarct dementia of Swedish type is caused by a 3'UTR mutation of COL4A1. *Brain*, **2017**, 140, e29	11.2	12
76	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimer's disease resilience. *Genome Medicine*, **2017**, 9, 100	14.4	40
75	Current concepts and controversies in the pathogenesis of Parkinson's disease dementia and Dementia with Lewy Bodies. *F1000Research*, **2017**, 6, 1604	3.6	23
74	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
73	Mutations in TYROBP are not a common cause of dementia in a Turkish cohort. *Neurobiology of Aging*, **2017**, 58, 240.e1-240.e3	5.6	5
72	[P3□01]: MULTI-INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3□UTR COL4A1 MUTATION **2017**, 13, P973-P973		
71	Study protocol: Insight 46 - a neuroscience sub-study of the MRC National Survey of Health and Development. *BMC Neurology*, **2017**, 17, 75	3.1	42
70	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
69	Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. *Neurobiology of Aging*, **2017**, 49, 215.e1-215.e8	5.6	15
68	[P3□10]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER'S DISEASE **2017**, 13, P976-P977		
67	[P3□11]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES **2017**, 13, P977-P977		
66	[P3□12]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER's DISEASE USING WHOLE-EXOME SEQUENCING **2017**, 13, P977-P977		

65	[F5□1□2]: GENETICS OF DLB AND RELEVANCE FOR MECHANISMS **2017**, 13, P1444		
64	[P4□216]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON TYROBP **2017**, 13, P1490-P1491		
63	Mutations in a Sibship with Multifocal Polymyoclonus. *Tremor and Other Hyperkinetic Movements*, **2017**, 7, 452	2	4
62	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. *Human Molecular Genetics*, **2016**, 25, 5483-5489	5.6	40
61	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. *Neurobiology of Aging*, **2016**, 46, 235.e1-9	5.6	33
60	The clinical syndrome of dystonia with anarthria/aphonia. *Parkinsonism and Related Disorders*, **2016**, 24, 20-7	3.6	7
59	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
58	Screening exons 16 and 17 of the amyloid precursor protein gene in sporadic early-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 39, 220.e1-7	5.6	9
57	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. *PLoS Medicine*, **2016**, 13, e1001976	11.6	100
56	Influence of Coding Variability in APP-A□Metabolism Genes in Sporadic Alzheimer's Disease. *PLoS ONE*, **2016**, 11, e0150079	3.7	26
55	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. *PLoS ONE*, **2016**, 11, e0162592	3.7	16
54	The Chihuahua dog: A new animal model for neuronal ceroid lipofuscinosis CLN7 disease?. *Journal of Neuroscience Research*, **2016**, 94, 339-47	4.4	22
53	Exome sequencing in a consanguineous family clinically diagnosed with early-onset Alzheimer's disease identifies a homozygous CTSF mutation. *Neurobiology of Aging*, **2016**, 46, 236.e1-6	5.6	19
52	Mutation of TBCK causes a rare recessive developmental disorder. *Neurology: Genetics*, **2016**, 2, e76	3.8	13
51	P2-158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK **2016**, 12, P675-P675		
50	P3-091: Investigating SARM1 Variants in Alzheimer□ Disease Cohorts **2016**, 12, P855-P855		
49	Genetic risk factors for the posterior cortical atrophy variant of Alzheimer's disease. *Alzheimer's and Dementia*, **2016**, 12, 862-71	1.2	64
48	TYROBP genetic variants in early-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 48, 222.e9-222.e15	5.6	51

47	RARS2 mutations in a sibship with infantile spasms. *Epilepsia*, **2016**, 57, e97-e102	6.4	18
46	Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. *American Journal of Human Genetics*, **2015**, 96, 474-9	11	90
45	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. *Neurology: Genetics*, **2015**, 1, e9	3.8	63
44	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. *Neurobiology of Aging*, **2015**, 36, 1605.e7-12	5.6	70
43	Genetic Variants and Related Biomarkers in Sporadic Alzheimer's Disease. *Current Genetic Medicine Reports*, **2015**, 3, 19-25	2.2	1
42	A systematic screening to identify de novo mutations causing sporadic early-onset Parkinson's disease. *Human Molecular Genetics*, **2015**, 24, 6711-20	5.6	26
41	The age factor in Alzheimer's disease. *Genome Medicine*, **2015**, 7, 106	14.4	137
40	Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. *International Journal of Molecular Sciences*, **2015**, 16, 24629-55	6.3	19
39	SnapShot: Genetics of ALS and FTD. *Cell*, **2015**, 160, 798-798.e1	56.2	54
38	SnapShot: Genetics of Parkinson's disease. *Cell*, **2015**, 160, 570-570.e1	56.2	67
37	A phenotype of atypical apraxia of speech in a family carrying SQSTM1 mutation. *Journal of Alzheimer's Disease*, **2015**, 43, 625-30	4.3	10
36	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. *Human Molecular Genetics*, **2014**, 23, R47-53	5.6	48
35	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. *Nature*, **2014** , 505, 550-554	50.4	345
34	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. *Nature Genetics*, **2014**, 46, 989-93	36.3	1261
33	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. *Brain*, **2014**, 137, 2480-92	11.2	127
32	Assessment of Parkinson's disease risk loci in Greece. *Neurobiology of Aging*, **2014**, 35, 442.e9-442.e16	5.6	12
31	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. *Neurobiology of Aging*, **2014**, 35, 2419.e23-2419.e25	5.6	63
30	Missense variant in TREML2 protects against Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 1510.e19-1510.e20	5.6	84

29	Nonsense mutation in PRNP associated with clinical Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 2656.e13-2656.e16	5.6	22
28	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
27	Atypical Parkinsonism-Dystonia Syndrome Caused by a Novel DJ1 Mutation. *Movement Disorders Clinical Practice*, **2014**, 1, 45-49	2.2	6
26	Investigating the role of rare coding variability in Mendelian dementia genes (APP, PSEN1, PSEN2, GRN, MAPT, and PRNP) in late-onset Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 2881.e1-2881.e6	5.6	45
25	Influence of single nucleotide polymorphisms in COMT, MAO-A and BDNF genes on dyskinesias and levodopa use in Parkinson's disease. *Neurodegenerative Diseases*, **2014**, 13, 24-8	2.3	40
24	SnapShot: genetics of Alzheimer's disease. *Cell*, **2013**, 155, 968-968.e1	56.2	78
23	Insights into TREM2 biology by network analysis of human brain gene expression data. *Neurobiology of Aging*, **2013**, 34, 2699-714	5.6	119
22	TREM2 variants in Alzheimer's disease. *New England Journal of Medicine*, **2013**, 368, 117-27	59.2	1805
21	CLN6 disease caused by the same mutation originating in Pakistan has varying pathology. *European Journal of Paediatric Neurology*, **2013**, 17, 657-60	3.8	8
20	Novel compound heterozygous mutation in TREM2 found in a Turkish frontotemporal dementia-like family. *Neurobiology of Aging*, **2013**, 34, 2890.e1-5	5.6	90
19	Genetic analysis of inherited leukodystrophies: genotype-phenotype correlations in the CSF1R gene. *JAMA Neurology*, **2013**, 70, 875-882	17.2	58
18	SQSTM1 mutations in French patients with frontotemporal dementia or frontotemporal dementia with amyotrophic lateral sclerosis. *JAMA Neurology*, **2013**, 70, 1403-10	17.2	131
17	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. *JAMA Neurology*, **2013**, 70, 727-35	17.2	285
16	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. *Movement Disorders*, **2012**, 27, 526-32	7	80
15	Tremor-ataxia with central hypomyelination (TACH): dystonia as a new clinical feature. *Movement Disorders*, **2012**, 27, 1829-30	7	23
14	Mutation of the parkinsonism gene ATP13A2 causes neuronal ceroid-lipofuscinosis. *Human Molecular Genetics*, **2012**, 21, 2646-50	5.6	196
13	Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. *Nature Reviews Neuroscience*, **2012**, 13, 453-64	13.5	89
12	Mutations in ANO3 cause dominant craniocervical dystonia: ion channel implicated in pathogenesis. *American Journal of Human Genetics*, **2012**, 91, 1041-50	11	172

11	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. *Human Molecular Genetics*, **2012**, 21, 4996-5009	5.6	145
10	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. *Brain*, **2009**, 132, 1783-94	11.2	488
9	SNCA variants are associated with increased risk for multiple system atrophy. *Annals of Neurology*, **2009**, 65, 610-4	9.4	232
8	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. *Neurobiology of Aging*, **2009**, 30, 1515-7	5.6	79
7	Analysis of Parkinson disease patients from Portugal for mutations in SNCA, PRKN, PINK1 and LRRK2. *BMC Neurology*, **2008**, 8, 1	3.1	32
6	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. *Lancet Neurology, The*, **2008**, 7, 207-15	24.1	159
5	Emerging pathways in genetic Parkinson's disease: Potential role of ceramide metabolism in Lewy body disease. *FEBS Journal*, **2008**, 275, 5767-73	5.7	109
4	How understudied populations have contributed to our understanding of Alzheimer's disease genetics		1
3	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer's Disease		1
2	A comprehensive assessment of benign genetic variability for neurodegenerative disorders		7
1	Expanding Parkinson's disease genetics: novel risk loci, genomic context, causal insights and heritable risk		51