Rita Guerreiro

List of Publications by Year in Descending Order

162 papers	15,517 citations	50 h-index	124 g-index
177 ext. papers	19,607 ext. citations	10.2 avg, IF	5.58 L-index

#	Paper	IF	Citations
162	Prion-like □synuclein pathology in the brain of infants with Krabbe disease.. *Brain*, **2022**,	11.2	2
161	Challenge accepted: uncovering the role of rare genetic variants in Alzheimer's disease.. *Molecular Neurodegeneration*, **2022**, 17, 3	19	1
160	Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank.. *Scientific Reports*, **2022**, 12, 8404	4.9	0
159	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. *Alzheimerls and Dementia*, **2021**,	1.2	2
158	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum NfL and pNfH: A Longitudinal Multicentre Study. *Annals of Neurology*, **2021**,	9.4	2
157	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. *Molecular Neurodegeneration*, **2021**, 16, 79	19	0
156	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. *Brain*, **2021**,	11.2	3
155	PHACTR1 genetic variability is not critical in small vessel ischemic disease patients and PcomA recruitment in C57BL/6J mice. *Scientific Reports*, **2021**, 11, 6072	4.9	1
154	How understudied populations have contributed to our understanding of Alzheimer's disease genetics. *Brain*, **2021**, 144, 1067-1081	11.2	2
153	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
152	Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. *Neuromuscular Disorders*, **2021**, 31, 891-895	2.9	
151	TREM2 variants as a possible cause of frontotemporal dementia with distinct neuroimaging features. *European Journal of Neurology*, **2021**, 28, 2603-2613	6	1
150	Genetics of synucleins in neurodegenerative diseases. *Acta Neuropathologica*, **2021**, 141, 471-490	14.3	6
149	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. *European Journal of Human Genetics*, **2021**, 29, 663-671	5.3	0
148	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. *Alzheimerls and Dementia*, **2021**, 17, 500-514	1.2	8
147	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
146	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

145	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. *JAMA Network Open*, **2021**, 4, e2030194	10.4	14
144	A comprehensive analysis of copy number variation in a Turkish dementia cohort. *Human Genomics*, **2021**, 15, 48	6.8	
143	Disease-related cortical thinning in presymptomatic granulin mutation carriers. *NeuroImage: Clinical* , **2021**, 29, 102540	5.3	2
142	Polygenic risk scores for Alzheimer's disease are related to dementia risk in APOE ε4 negatives. *Alzheimerls and Dementia: Diagnosis, Assessment and Disease Monitoring*, **2021**, 13, e12142	5.2	5
141	Analysis of copy number variation in a Turkish dementia cohort. *Alzheimerls and Dementia*, **2020**, 16, e044868	12.8	1
140	KCNN2 mutation in autosomal-dominant tremulous myoclonus-dystonia. *European Journal of Neurology*, **2020**, 27, 1471-1477	6	10
139	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
138	Alzheimer's Disease Genetics: Review of Novel Loci Associated with Disease. *Current Genetic Medicine Reports*, **2020**, 8, 1-16	2.2	12
137	Novel Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. *Journal of Clinical Medicine*, **2020**, 9,	5.1	1
136	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. *Acta Neuropathologica Communications*, **2020**, 8, 5	7.3	15
135	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. *Cortex*, **2020**, 133, 384-398	3.8	7
134	Genetic architecture of common non-Alzheimer's disease dementias. *Neurobiology of Disease*, **2020**, 142, 104946	7.5	16
133	A locked immunometabolic switch underlies TREM2 R47H loss of function in human iPSC-derived microglia. *FASEB Journal*, **2020**, 34, 2436-2450	0.9	35
132	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
131	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
130	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
129	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
128	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. *Brain Communications*, **2020**, 2,	4.5	6

127	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
126	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
125	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
124	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. *Movement Disorders*, **2019**, 34, 460-468	7	40
123	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. *NeuroImage*, **2019**, 189, 645-654	7.9	18
122	Alzheimer's disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. *Translational Psychiatry*, **2019**, 9, 154	8.6	31
121	Heritability and genetic variance of dementia with Lewy bodies. *Neurobiology of Disease*, **2019**, 127, 492-501	7.9	15
120	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
119	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. *Lancet Neurology, The*, **2019**, 18, 1103-1111	24.1	68
118	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
117	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. *Acta Neuropathologica Communications*, **2019**, 7, 188	7.3	6
116	Is APOE □4 required for Alzheimer's disease to develop in TREM2 p.R47H variant carriers?. *Neuropathology and Applied Neurobiology*, **2019**, 45, 187-189	5.2	2
115	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 Alzheimerls Disease*, **2019**, 67, 159-167	4.3	7
114	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. *Neurobiology of Aging*, **2019**, 77, 169-177	5.6	24
113	A comprehensive screening of copy number variability in dementia with Lewy bodies. *Neurobiology of Aging*, **2019**, 75, 223.e1-223.e10	5.6	10
112	Genetics of dementia in a Finnish cohort. *European Journal of Human Genetics*, **2018**, 26, 827-837	5.3	5
111	The genetic landscape of Alzheimer disease. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2018**, 148, 395-408	3	56
110	Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. *Neurology: Genetics*, **2018**, 4, e215	3.8	8

109	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
108	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. *Neurobiology of Aging*, **2018**, 62, 244.e1-244.e8	5.6	25
107	Genotyping of the Alzheimer's Disease Genome-Wide Association Study Index Single Nucleotide Polymorphisms in the Brains for Dementia Research Cohort. *Journal of Alzheimerls Disease*, **2018**, 64, 355-362	4.3	6
106	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. *JAMA Neurology*, **2018**, 75, 1416-1422	17.2	50
105	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
104	The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. *Lancet Neurology, The*, **2018**, 17, 721-730	24.1	92
103	The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. *Current Neurology and Neuroscience Reports*, **2018**, 18, 67	6.6	41
102	Whole-exome sequencing of the BDR cohort: evidence to support the role of the PILRA gene in Alzheimer's disease. *Neuropathology and Applied Neurobiology*, **2018**, 44, 506-521	5.2	27
101	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
100	LRP10 in □synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032	24.1	14
99	Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddon's Syndrome of Unknown Cause. *Cerebrovascular Diseases*, **2018**, 46, 257-264	3.2	10
98	LRP10 in □synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032-1033	24.1	9
97	splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. *Neurology: Genetics*, **2018**, 4, e273	3.8	4
96	Genetically elevated high-density lipoprotein cholesterol through the cholesteryl ester transfer protein gene does not associate with risk of Alzheimer's disease. *Alzheimerls and Dementia: Diagnosis, Assessment and Disease Monitoring*, **2018**, 10, 595-598	5.2	
95	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
94	Multi-infarct dementia of Swedish type is caused by a 3'UTR mutation of COL4A1. *Brain*, **2017**, 140, e29	11.2	12
93	A recessive ataxia diagnosis algorithm for the next generation sequencing era. *Annals of Neurology*, **2017**, 82, 892-899	9.4	20
92	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimer's disease resilience. *Genome Medicine*, **2017**, 9, 100	14.4	40

91	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
90	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
89	[P3□101]: MULTI-INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3□UTR COL4A1 MUTATION **2017**, 13, P973-P973		
88	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
87	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
86	Primary familial brain calcification linked to deletion of 5' noncoding region of SLC20A2. *Acta Neurologica Scandinavica*, **2017**, 136, 59-63	3.8	6
85	[P3□110]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER's DISEASE **2017**, 13, P976-P977		
84	[P3□111]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES **2017**, 13, P977-P977		
83	[P3□112]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER's DISEASE USING WHOLE-EXOME SEQUENCING **2017**, 13, P977-P977		
82	[P4□216]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON TYROBP **2017**, 13, P1490-P1491		
81	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. *Brain*, **2017**, 140, 3191-3203	11.2	209
80	Mutations in a Sibship with Multifocal Polymyoclonus. *Tremor and Other Hyperkinetic Movements*, **2017**, 7, 452	2	4
79	Pseudohypoparathyroidism type I-b with neurological involvement is associated with a homozygous PTH1R mutation. *Genes, Brain and Behavior*, **2016**, 15, 669-77	3.6	7
78	Clinical phenotype and genetic associations in autosomal dominant familial Alzheimer's disease: a case series. *Lancet Neurology, The*, **2016**, 15, 1326-1335	24.1	109
77	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. *Neurobiology of Aging*, **2016**, 46, 235.e1-9	5.6	33
76	The clinical syndrome of dystonia with anarthria/aphonia. *Parkinsonism and Related Disorders*, **2016**, 24, 20-7	3.6	7
75	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
74	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

73	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. *American Journal of Human Genetics*, **2016**, 98, 500-513	11	225
72	Influence of Coding Variability in APP-A Metabolism Genes in Sporadic Alzheimer's Disease. *PLoS ONE*, **2016**, 11, e0150079	3.7	26
71	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
70	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
69	P2-158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK **2016**, 12, P675-P675		
68	P3-091: Investigating SARM1 Variants in Alzheimer Disease Cohorts **2016**, 12, P855-P855		
67	Characterization of an FTLD-PDB family with the coexistence of SQSTM1 mutation and hexanucleotide (GC repeat expansion in C9orf72 gene. *Neurobiology of Aging*, **2016**, 40, 191.e1-191.e8	5.6	9
66	TYROBP genetic variants in early-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 48, 222.e9-222.e15	5.6	51
65	RARS2 mutations in a sibship with infantile spasms. *Epilepsia*, **2016**, 57, e97-e102	6.4	18
64	Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. *American Journal of Human Genetics*, **2015**, 96, 474-9	11	90
63	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. *Neurology: Genetics*, **2015**, 1, e9	3.8	63
62	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. *Neurobiology of Aging*, **2015**, 36, 1605.e7-12	5.6	70
61	Genetic Variants and Related Biomarkers in Sporadic Alzheimer's Disease. *Current Genetic Medicine Reports*, **2015**, 3, 19-25	2.2	1
60	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
59	A Novel MAPT Mutation Causing Corticobasal Syndrome Led by Progressive Apraxia of Speech. *Journal of Alzheimer's Disease*, **2015**, 48, 923-6	4.3	12
58	The age factor in Alzheimer's disease. *Genome Medicine*, **2015**, 7, 106	14.4	137
57	SnapShot: Genetics of ALS and FTD. *Cell*, **2015**, 160, 798-798.e1	56.2	54
56	SnapShot: Genetics of Parkinson's disease. *Cell*, **2015**, 160, 570-570.e1	56.2	67

55	A phenotype of atypical apraxia of speech in a family carrying SQSTM1 mutation. *Journal of Alzheimerls Disease*, **2015**, 43, 625-30	4.3	10
54	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. *Human Molecular Genetics*, **2014**, 23, R47-53	5.6	48
53	Exome sequencing identifies 2 novel presenilin 1 mutations (p.L166V and p.S230R) in British early-onset Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 2422.e13-6	5.6	26
52	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. *Nature*, **2014** , 505, 550-554	50.4	345
51	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. *Neurobiology of Aging*, **2014**, 35, 2419.e23-2419.e25	5.6	63
50	Missense variant in TREML2 protects against Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 1510.e19-1510.e26	5.6	84
49	Nonsense mutation in PRNP associated with clinical Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 2656.e13-2656.e16	5.6	22
48	Unbiased screen for interactors of leucine-rich repeat kinase 2 supports a common pathway for sporadic and familial Parkinson disease. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 2626-31	11.5	282
47	Genetics of Alzheimer's disease. *Neurotherapeutics*, **2014**, 11, 732-7	6.4	108
46	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
45	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
44	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. *Human Molecular Genetics*, **2014**, 23, 562-562	5.6	3
43	Analysis of genome-wide association studies of Alzheimer disease and of Parkinson disease to determine if these 2 diseases share a common genetic risk. *JAMA Neurology*, **2013**, 70, 1268-76	17.2	46
42	A paired RNAi and RabGAP overexpression screen identifies Rab11 as a regulator of β-amyloid production. *Cell Reports*, **2013**, 5, 1536-51	10.6	84
41	SnapShot: genetics of Alzheimer's disease. *Cell*, **2013**, 155, 968-968.e1	56.2	78
40	Insights into TREM2 biology by network analysis of human brain gene expression data. *Neurobiology of Aging*, **2013**, 34, 2699-714	5.6	119
39	TREM2 variants in Alzheimer's disease. *New England Journal of Medicine*, **2013**, 368, 117-27	59.2	1805
38	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

37	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
36	A novel A781V mutation in the CSF1R gene causes hereditary diffuse leucoencephalopathy with axonal spheroids. *Journal of the Neurological Sciences*, **2013**, 332, 141-4	3.2	14
35	Genetic analysis of inherited leukodystrophies: genotype-phenotype correlations in the CSF1R gene. *JAMA Neurology*, **2013**, 70, 875-882	17.2	58
34	Using exome sequencing to reveal mutations in TREM2 presenting as a frontotemporal dementia-like syndrome without bone involvement. *JAMA Neurology*, **2013**, 70, 78-84	17.2	257
33	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. *Human Molecular Genetics*, **2013**, 22, 1039-49	5.6	96
32	SQSTM1 mutations in French patients with frontotemporal dementia or frontotemporal dementia with amyotrophic lateral sclerosis. *JAMA Neurology*, **2013**, 70, 1403-10	17.2	131
31	Initial assessment of the pathogenic mechanisms of the recently identified Alzheimer risk Loci. *Annals of Human Genetics*, **2013**, 77, 85-105	2.2	40
30	The Val158Met COMT polymorphism is a modifier of the age at onset in Parkinson's disease with a sexual dimorphism. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, 666-73	5.5	40
29	NOTCH3 variants and risk of ischemic stroke. *PLoS ONE*, **2013**, 8, e75035	3.7	27
28	Duplication of amyloid precursor protein (APP), but not prion protein (PRNP) gene is a significant cause of early onset dementia in a large UK series. *Neurobiology of Aging*, **2012**, 33, 426.e13-21	5.6	55
27	Screening for VPS35 mutations in Parkinson's disease. *Neurobiology of Aging*, **2012**, 33, 838.e1-5	5.6	45
26	Tau acts as an independent genetic risk factor in pathologically proven PD. *Neurobiology of Aging*, **2012**, 33, 838.e7-11	5.6	20
25	Genetic testing in familial and young-onset Alzheimer's disease: mutation spectrum in a Serbian cohort. *Neurobiology of Aging*, **2012**, 33, 1481.e7-12	5.6	15
24	Complement receptor 1 (CR1) and Alzheimer's disease. *Immunobiology*, **2012**, 217, 244-50	3.4	95
23	Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. *Nature Reviews Neuroscience*, **2012**, 13, 453-64	13.5	89
22	Identification of Stk25 as a genetic modifier of Tau phosphorylation in Dab1-mutant mice. *PLoS ONE*, **2012**, 7, e31152	3.7	10
21	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. *Human Molecular Genetics*, **2012**, 21, 4996-5009	5.6	145
20	The role of variation at APP, PSEN1, PSEN2, and MAPT in late onset Alzheimer's disease. *Journal of Alzheimerls Disease*, **2012**, 28, 377-87	4.3	47

19	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. *Nature Genetics*, **2011**, 43, 429-35	36.3	1421
18	Familial frontotemporal dementia with amyotrophic lateral sclerosis and a shared haplotype on chromosome 9p. *Journal of Neurology*, **2011**, 258, 647-55	5.5	71
17	A new way APP mismetabolism can lead to Alzheimer's disease. *EMBO Molecular Medicine*, **2011**, 3, 247-8	8.2	3
16	Clusterin as an Alzheimer biomarker. *Archives of Neurology*, **2011**, 68, 1459-60		8
15	Genetic evidence implicates the immune system and cholesterol metabolism in the aetiology of Alzheimer's disease. *PLoS ONE*, **2010**, 5, e13950	3.7	276
14	PRNP allelic series from 19 years of prion protein gene sequencing at the MRC Prion Unit. *Human Mutation*, **2010**, 31, E1551-63	4.7	70
13	The heritability and genetics of frontotemporal lobar degeneration. *Neurology*, **2009**, 73, 1451-6	6.5	339
12	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. *Brain*, **2009**, 132, 1783-94	11.2	488
11	Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. *Nature Genetics*, **2009**, 41, 1088-93	36.3	2018
10	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. *Neurobiology of Aging*, **2009**, 30, 1515-7	5.6	79
9	Ubiquitin associated protein 1 is a risk factor for frontotemporal lobar degeneration. *Neurobiology of Aging*, **2009**, 30, 656-65	5.6	29
8	Genotype, haplotype and copy-number variation in worldwide human populations. *Nature*, **2008**, 451, 998-1003	50.4	662
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	How understudied populations have contributed to our understanding of Alzheimer's disease genetics		1
4	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer's Disease		1
3	A comprehensive assessment of benign genetic variability for neurodegenerative disorders		7
2	Meta-analysis of genetic association with diagnosed Alzheimer's disease identifies novel risk loci and implicates Abeta, Tau, immunity and lipid processing		9

List of Publications

1 A locked immunometabolic switch underlies TREM2 R47H loss of function in human iPSC--derived microglia 1