Rita Guerreiro

List of Publications by Year
in descending order

163 papers	22,763 citations	29994 54 h-index	9839 141 g-index
177 all docs	177 docs citations	177 times ranked	25801 citing authors

#	Article	IF	Citations
1	Genome-wide association study identifies variants at CLU and PICALM associated with Alzheimer's disease. Nature Genetics, 2009, 41, 1088-1093.	9.4	2,697
2	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	13.9	2,385
3	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
4	Common variants at ABCA7, MS4A6A/MS4A4E, EPHA1, CD33 and CD2AP are associated with Alzheimer's disease. Nature Genetics, 2011, 43, 429-435.	9.4	1,708
5	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.	4.9	1,414
6	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
7	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	9.4	783
8	Genotype, haplotype and copy-number variation in worldwide human populations. Nature, 2008, 451, 998-1003.	13.7	780
9	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. Brain, 2009, 132, 1783-1794.	3.7	612
10	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	13.7	425
11	The heritability and genetics of frontotemporal lobar degeneration. Neurology, 2009, 73, 1451-1456.	1.5	416
12	Genetic Evidence Implicates the Immune System and Cholesterol Metabolism in the Aetiology of Alzheimer's Disease. PLoS ONE, 2010, 5, e13950.	1.1	347
13	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, 2626-2631.	3.3	342
14	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.	2.6	333
15	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
16	Using Exome Sequencing to Reveal Mutations in TREM2 Presenting as a Frontotemporal Dementiaâ€“like Syndrome Without Bone Involvement. JAMA Neurology, 2013, 70, 78.	4.5	311
17	The age factor in Alzheimerâ€™s disease. Genome Medicine, 2015, 7, 106.	3.6	271
18	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. Lancet Neurology, The, 2008, 7, 207-215.	4.9	202

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19	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. Lancet Neurology, The, 2018, 17, 64-74.	4.9	195
20	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. Human Molecular Genetics, 2014, 23, 6139-6146.	1.4	178
21	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
22	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.	4.9	175
23	Clinical phenotype and genetic associations in autosomal dominant familial Alzheimerâ€™s disease: a case series. Lancet Neurology, The, 2016, 15, 1326-1335.	4.9	163
24	The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. Lancet Neurology, The, 2018, 17, 721-730.	4.9	161
25	SQSTM1Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. JAMA Neurology, 2013, 70, 1403-10.	4.5	153
26	Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875.	4.5	147
27	Insights into TREM2 biology by network analysis of human brain gene expression data. Neurobiology of Aging, 2013, 34, 2699-2714.	1.5	145
28	Genetics of Alzheimer's Disease. Neurotherapeutics, 2014, 11, 732-737.	2.1	134
29	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. Lancet Neurology, The, 2019, 18, 1103-1111.	4.9	128
30	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	2.6	127
31	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
32	A Paired RNAi and RabGAP Overexpression Screen Identifies Rab11 as a Regulator of Î²-Amyloid Production. Cell Reports, 2013, 5, 1536-1551.	2.9	120
33	A novel compound heterozygous mutation in TREM2 found in a Turkish frontotemporal dementia-like family. Neurobiology of Aging, 2013, 34, 2890.e1-2890.e5.	1.5	113
34	Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. Nature Reviews Neuroscience, 2012, 13, 453-464.	4.9	110
35	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	1.5	110
36	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.	1.5	108

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37	Complement receptor 1 (CR1) and Alzheimer's disease. Immunobiology, 2012, 217, 244-250.	0.8	107
38	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. Neurobiology of Aging, 2009, 30, 1515-1517.	1.5	97
39	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
40	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	4.5	95
41	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. Neurology: Genetics, 2015, 1, e9.	0.9	90
42	SnapShot: Genetics of Alzheimerâ€™s Disease. Cell, 2013, 155, 968-968.e1.	13.5	86
43	The genetic landscape of Alzheimer disease. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 395-408.	1.0	86
44	PRNP allelic series from 19 years of prion protein gene sequencing at the MRC Prion Unit. Human Mutation, 2010, 31, E1551-E1563.	1.1	85
45	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. Neurobiology of Aging, 2014, 35, 2419.e23-2419.e25.	1.5	84
46	A locked immunometabolic switch underlies TREM2 R47H loss of function in human iPSCâ€derived microglia. FASEB Journal, 2020, 34, 2436-2450.	0.2	82
47	SnapShot: Genetics of Parkinsonâ€™s Disease. Cell, 2015, 160, 570-570.e1.	13.5	79
48	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.	1.5	78
49	Familial frontotemporal dementia with amyotrophic lateral sclerosis and a shared haplotype on chromosomeÂ9p. Journal of Neurology, 2011, 258, 647-655.	1.8	76
50	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	1.5	69
51	The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. Current Neurology and Neuroscience Reports, 2018, 18, 67.	2.0	69
52	Alzheimerâ€™s disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. Translational Psychiatry, 2019, 9, 154.	2.4	69
53	SnapShot: Genetics of ALS and FTD. Cell, 2015, 160, 798-798.e1.	13.5	68
54	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.e13-426.e21.	1.5	67

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55	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimerâ€™s disease resilience. Genome Medicine, 2017, 9, 100.	3.6	67
56	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	4.5	66
57	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	2.2	66
58	A systematic screening to identifyde novomutations causing sporadic early-onset Parkinson's disease. Human Molecular Genetics, 2015, 24, 6711-6720.	1.4	59
59	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. Human Molecular Genetics, 2014, 23, R47-R53.	1.4	57
60	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	2.2	57
61	The Role of Variation at AÎ²PP, PSEN1, PSEN2, and MAPT in Late Onset Alzheimer's Disease. Journal of Alzheimer's Disease, 2012, 28, 377-387.	1.2	53
62	Screening for VPS35 mutations in Parkinson's disease. Neurobiology of Aging, 2012, 33, 838.e1-838.e5.	1.5	53
63	Investigating the role of rare coding variability in Mendelian dementia genes (APP , PSEN1 , PSEN2 , GRN) Tj ETQq1 1 0.784314 rgBT /O	1.5	53
64	Analysis of Parkinson disease patients from Portugal for mutations in SNCA, PRKN, PINK1 and LRRK2. BMC Neurology, 2008, 8, 1.	0.8	52
65	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.	4.5	51
66	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	2.2	47
67	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. Neurobiology of Aging, 2019, 77, 169-177.	1.5	47
68	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, 666-673.	0.9	43
69	A rare loss-of-function variant of ADAM17 is associated with late-onset familial Alzheimer disease. Molecular Psychiatry, 2020, 25, 629-639.	4.1	42
70	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. JAMA Network Open, 2021, 4, e2030194.	2.8	42
71	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105.	0.3	41
72	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. Neurobiology of Aging, 2016, 46, 235.e1-235.e9.	1.5	37

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73	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. Alzheimer's and Dementia, 2021, 17, 500-514.	0.4	36
74	Wholeâ€exome sequencing of the <scp>BDR</scp> cohort: evidence to support the role of the <scp>PILRA</scp> gene in Alzheimer's disease. Neuropathology and Applied Neurobiology, 2018, 44, 506-521.	1.8	35
75	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.e1-236.e6.	1.5	34
76	Influence of Coding Variability in APP-AÎ² Metabolism Genes in Sporadic Alzheimerâ€™s Disease. PLoS ONE, 2016, 11, e0150079.	1.1	34
77	Ubiquitin associated protein 1 is a risk factor for frontotemporal lobar degeneration. Neurobiology of Aging, 2009, 30, 656-665.	1.5	33
78	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. NeuroImage, 2019, 189, 645-654.	2.1	33
79	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. Neurobiology of Aging, 2018, 66, 179.e17-179.e29.	1.5	32
80	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. Neurobiology of Aging, 2018, 62, 244.e1-244.e8.	1.5	30
81	NOTCH3 Variants and Risk of Ischemic Stroke. PLoS ONE, 2013, 8, e75035.	1.1	30
82	Heritability and genetic variance of dementia with Lewy bodies. Neurobiology of Disease, 2019, 127, 492-501.	2.1	29
83	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.e13-2422.e16.	1.5	28
84	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	2.8	27
85	Genetic architecture of common non-Alzheimerâ€™s disease dementias. Neurobiology of Disease, 2020, 142, 104946.	2.1	27
86	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. Acta Neuropathologica Communications, 2020, 8, 5.	2.4	27
87	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. Brain, 2022, 145, 1805-1817.	3.7	27
88	Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank. Scientific Reports, 2022, 12, 8404.	1.6	27
89	A nonsense mutation in PRNP associated with clinical Alzheimer's disease. Neurobiology of Aging, 2014, 35, 2656.e13-2656.e16.	1.5	26
90	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. Cortex, 2020, 133, 384-398.	1.1	26

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91	Polygenic risk scores for Alzheimer's disease are related to dementia risk in APOE É›4 negatives. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12142.	1.2	25
92	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. Alzheimer's and Dementia, 2022, 18, 1408-1423.	0.4	24
93	Tau acts as an independent genetic risk factor in pathologically proven PD. Neurobiology of Aging, 2012, 33, 838.e7-838.e11.	1.5	23
94	<scp>RARS</scp>2 mutations in a sibship with infantile spasms. Epilepsia, 2016, 57, e97-e102.	2.6	23
95	Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. Neurobiology of Aging, 2017, 49, 215.e1-215.e8.	1.5	21
96	KCNN2 mutation in autosomalâ€dominant tremulous myoclonusâ€dystonia. European Journal of Neurology, 2020, 27, 1471-1477.	1.7	21
97	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum <scp>NfL</scp> and <scp>pNfH</scp>: A Longitudinal Multicentre Study. Annals of Neurology, 2022, 91, 33-47.	2.8	21
98	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. Brain Communications, 2020, 2, .	1.5	20
99	Alzheimerâ€™s Disease Genetics: Review of Novel Loci Associated with Disease. Current Genetic Medicine Reports, 2020, 8, 1-16.	1.9	20
100	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. PLoS ONE, 2016, 11, e0162592.	1.1	19
101	Multi-infarct dementia of Swedish type is caused by a 3â€™UTR mutation of COL4A1. Brain, 2017, 140, e29-e29.	3.7	19
102	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. Acta Neuropathologica Communications, 2019, 7, 188.	2.4	19
103	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. Annals of Neurology, 2020, 88, 113-122.	2.8	19
104	Challenge accepted: uncovering the role of rare genetic variants in Alzheimerâ€™s disease. Molecular Neurodegeneration, 2022, 17, 3.	4.4	19
105	Genetic testing in familial and young-onset Alzheimer's disease: mutation spectrum in a Serbian cohort. Neurobiology of Aging, 2012, 33, 1481.e7-1481.e12.	1.5	18
106	A novel A781V mutation in the CSF1R gene causes hereditary diffuse leucoencephalopathy with axonal spheroids. Journal of the Neurological Sciences, 2013, 332, 141-144.	0.3	18
107	A Non-APOE Polygenic Risk Score for Alzheimerâ€™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.	1.7	18
108	CLN6 disease caused by the same mutation originating in Pakistan has varying pathology. European Journal of Paediatric Neurology, 2013, 17, 657-660.	0.7	17

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109	A Novel MAPT Mutation Causing Corticobasal Syndrome Led by Progressive Apraxia of Speech. Journal of Alzheimer's Disease, 2015, 48, 923-926.	1.2	16
110	Genetics of synucleins in neurodegenerative diseases. Acta Neuropathologica, 2021, 141, 471-490.	3.9	16
111	Identification of Stk25 as a Genetic Modifier of Tau Phosphorylation in Dab1-Mutant Mice. PLoS ONE, 2012, 7, e31152.	1.1	15
112	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	4.9	15
113	Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddonâ€™s Syndrome of Unknown Cause. Cerebrovascular Diseases, 2018, 46, 257-264.	0.8	15
114	A comprehensive screening of copy number variability in dementia with Lewy bodies. Neurobiology of Aging, 2019, 75, 223.e1-223.e10.	1.5	13
115	A Phenotype of Atypical Apraxia of Speech in a Family Carrying SQSTM1 Mutation. Journal of Alzheimer's Disease, 2014, 43, 625-630.	1.2	12
116	Pseudohypoparathyroidism type Iâ€b with neurological involvement is associated with a homozygous <scp>PTH1R</scp> mutation. Genes, Brain and Behavior, 2016, 15, 669-677.	1.1	12
117	Screening exons 16 and 17 of the amyloid precursor protein gene in sporadic early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 39, 220.e1-220.e7.	1.5	12
118	Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. Neurobiology of Aging, 2017, 49, 214.e13-214.e15.	1.5	12
119	Characterization of an FTLD-PDB family with the coexistence of SQSTM1 mutation and hexanucleotide (G 4 C 2) repeat expansion inÂ C9orf72 gene. Neurobiology of Aging, 2016, 40, 191.e1-191.e8.	1.5	11
120	Primary familial brain calcification linked to deletion of 5â€™ noncoding region of SLC20A2. Acta Neurologica Scandinavica, 2017, 136, 59-63.	1.0	11
121	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032-1033.	4.9	11
122	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.	1.2	11
123	The clinical syndrome of dystonia with anarthria/aphonia. Parkinsonism and Related Disorders, 2016, 24, 20-27.	1.1	10
124	Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. Neurology: Genetics, 2018, 4, e215.	0.9	10
125	How understudied populations have contributed to our understanding of Alzheimerâ€™s disease genetics. Brain, 2021, 144, 1067-1081.	3.7	10
126	Clusterin as an Alzheimer Biomarker. Archives of Neurology, 2011, 68, 1459.	4.9	9

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127	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. Molecular Neurodegeneration, 2021, 16, 79.	4.4	9
128	Prion-like Î±-synuclein pathology in the brain of infants with Krabbe disease. Brain, 2022, 145, 1257-1263.	3.7	9
129	Disease-related cortical thinning in presymptomatic granulin mutation carriers. NeuroImage: Clinical, 2021, 29, 102540.	1.4	8
130	Genetic variants in glutamate-, AÎ²â^', and tau-related pathways determine polygenic risk for Alzheimer's disease. Neurobiology of Aging, 2021, 101, 299.e13-299.e21.	1.5	7
131	A new way APP mismetabolism can lead to Alzheimer's disease. EMBO Molecular Medicine, 2011, 3, 247-248.	3.3	6
132	Mutations in TYROBP are not a common cause of dementia in a Turkish cohort. Neurobiology of Aging, 2017, 58, 240.e1-240.e3.	1.5	6
133	Genetics of dementia in a Finnish cohort. European Journal of Human Genetics, 2018, 26, 827-837.	1.4	6
134	AP4S1 splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. Neurology: Genetics, 2018, 4, e273.	0.9	6
135	Genotyping of the Alzheimerâ€™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.	1.2	6
136	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. European Journal of Human Genetics, 2021, 29, 663-671.	1.4	6
137	Genetic analysis reveals novel variants for vascular cognitive impairment. Acta Neurologica Scandinavica, 2022, 146, 42-50.	1.0	6
138	Molecular Characterization of Portuguese Patients with Hereditary Cerebellar Ataxia. Cells, 2022, 11, 981.	1.8	6
139	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2014, 23, 562-562.	1.4	5
140	TREM2 variants as a possible cause of frontotemporal dementia with distinct neuroimaging features. European Journal of Neurology, 2021, 28, 2603-2613.	1.7	5
141	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.	0.6	4
142	Mutations in a Sibship with Multifocal Polymyoclonus. Tremor and Other Hyperkinetic Movements, 2017, 7, 452.	1.1	4
143	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2013, 22, 1696-1696.	1.4	3
144	Genetic Variants and Related Biomarkers in Sporadic Alzheimerâ€™s Disease. Current Genetic Medicine Reports, 2015, 3, 19-25.	1.9	3

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145	Two pathologically confirmed cases of novel mutations in the MAPT gene causing frontotemporal dementia. Neurobiology of Aging, 2020, 87, 141.e15-141.e20.	1.5	3
146	Patients with progranulin mutations overlap with the progressive dysexecutive syndrome: towards the definition of a frontoparietal dementia phenotype. Brain Communications, 2020, 2, fcaa126.	1.5	3
147	Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. Journal of Clinical Medicine, 2020, 9, 1212.	1.0	3
148	Genetically elevated highâ€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.	1.2	2
149	Is <scp>APOE</scp> Îµ4 required for Alzheimer's disease to develop in <scp>TREM</scp>2 p.R47H variant carriers?. Neuropathology and Applied Neurobiology, 2019, 45, 187-189.	1.8	2
150	PHACTR1 genetic variability is not critical in small vessel ischemic disease patients and PcomA recruitment in C57BL/6J mice. Scientific Reports, 2021, 11, 6072.	1.6	2
151	Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. Neuromuscular Disorders, 2021, 31, 891-895.	0.3	2
152	Exome Sequencing of a Portuguese Cohort of Frontotemporal Dementia Patients: Looking Into the ALS-FTD Continuum. Frontiers in Neurology, 0, 13, .	1.1	2
153	Analysis of copy number variation in a Turkish dementia cohort. Alzheimer's and Dementia, 2020, 16, e044868.	0.4	1
154	Rare variants in TP73 in a frontotemporal dementia cohort link this gene with primary progressive aphasia phenotypes. European Journal of Neurology, 2022, , .	1.7	1
155	Structural brain splitting is a hallmark of Granulin-related frontotemporal dementia. Neurobiology of Aging, 2022, , .	1.5	1
156	P2â€158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK. Alzheimer's and Dementia, 2016, 12, P675.	0.4	0
157	P3-091: Investigating SARM1 Variants in Alzheimerâ€™s Disease Cohorts. , 2016, 12, P855-P855.		0
158	[P3â€“101]: MULTIâ€INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3â€™UTR COL4A1 MUTATION. Alzheimer's and Dementia, 2017, 13, P973.	0.4	0
159	[P3â€“110]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER's DISEASE. Alzheimer's and Dementia, 2017, 13, P976.	0.4	0
160	[P3â€“111]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES. Alzheimer's and Dementia, 2017, 13, P977.	0.4	0
161	[P3â€“112]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER's DISEASE USING WHOLEâ€EXOME SEQUENCING. Alzheimer's and Dementia, 2017, 13, P977.	0.4	0
162	[P4â€“416]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON TYROBP. Alzheimer's and Dementia, 2017, 13, P1490.	0.4	0

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163	A comprehensive analysis of copy number variation in a Turkish dementia cohort. Human Genomics, 2021, 15, 48.	1.4	0