

Jose T Bras

List of Publications by Year in descending order

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Version: 2024-02-01

156
papers

20,872
citations

31949

53
h-index

11928

134
g-index

176
all docs

176
docs citations

176
times ranked

25390
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnosis and management of dementia with Lewy bodies. <i>Neurology</i> , 2017, 89, 88-100.	1.5	2,805
2	<i>TREM2</i> Variants in Alzheimer's Disease. <i>New England Journal of Medicine</i> , 2013, 368, 117-127.	13.9	2,385
3	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates β , tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019, 51, 414-430.	9.4	1,962
4	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. <i>Nature Genetics</i> , 2014, 46, 989-993.	9.4	1,685
5	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet Neurology</i> , The, 2019, 18, 1091-1102.	4.9	1,414
6	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
7	Rare coding variants in <i>PLCG2</i> , <i>ABI3</i> , and <i>TREM2</i> implicate microglial-mediated innate immunity in Alzheimer's disease. <i>Nature Genetics</i> , 2017, 49, 1373-1384.	9.4	783
8	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. <i>Brain</i> , 2009, 132, 1783-1794.	3.7	612
9	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. <i>Nature</i> , 2014, 505, 550-554.	13.7	425
10	A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. <i>JAMA Neurology</i> , 2013, 70, 727.	4.5	374
11	The age factor in Alzheimer's disease. <i>Genome Medicine</i> , 2015, 7, 106.	3.6	271
12	<i>SNCA</i> variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009, 65, 610-614.	2.8	257
13	Mutation of the parkinsonism gene <i>ATP13A2</i> causes neuronal ceroid-lipofuscinosis. <i>Human Molecular Genetics</i> , 2012, 21, 2646-2650.	1.4	231
14	Mutations in <i>ANO3</i> Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. <i>American Journal of Human Genetics</i> , 2012, 91, 1041-1050.	2.6	224
15	<i>DYT16</i> , a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein <i>PRKRA</i> . <i>Lancet Neurology</i> , The, 2008, 7, 207-215.	4.9	202
16	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. <i>Lancet Neurology</i> , The, 2018, 17, 64-74.	4.9	195
17	Genetic analysis implicates <i>APOE</i> , <i>SNCA</i> and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. <i>Human Molecular Genetics</i> , 2014, 23, 6139-6146.	1.4	178
18	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2012, 21, 4996-5009.	1.4	176

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19	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. <i>Lancet Neurology</i> , The, 2020, 19, 145-156.	4.9	175
20	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , 2014, 137, 2480-2492.	3.7	169
21	The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. <i>Lancet Neurology</i> , The, 2018, 17, 721-730.	4.9	161
22	<i>SQSTM1</i> Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2013, 70, 1403-10.	4.5	153
23	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. <i>PLoS Medicine</i> , 2016, 13, e1001976.	3.9	150
24	Genetic Analysis of Inherited Leukodystrophies. <i>JAMA Neurology</i> , 2013, 70, 875.	4.5	147
25	Insights into TREM2 biology by network analysis of human brain gene expression data. <i>Neurobiology of Aging</i> , 2013, 34, 2699-2714.	1.5	145
26	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. <i>Lancet Neurology</i> , The, 2019, 18, 1103-1111.	4.9	128
27	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. <i>American Journal of Human Genetics</i> , 2015, 96, 474-479.	2.6	127
28	Emerging pathways in genetic Parkinson's disease: Potential role of ceramide metabolism in Lewy body disease. <i>FEBS Journal</i> , 2008, 275, 5767-5773.	2.2	121
29	A novel compound heterozygous mutation in TREM2 found in a Turkish frontotemporal dementia-like family. <i>Neurobiology of Aging</i> , 2013, 34, 2890.e1-2890.e5.	1.5	113
30	Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. <i>Nature Reviews Neuroscience</i> , 2012, 13, 453-464.	4.9	110
31	Missense variant in TREML2 protects against Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014, 35, 1510.e19-1510.e26.	1.5	110
32	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. <i>Movement Disorders</i> , 2012, 27, 526-532.	2.2	108
33	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017, 57, 247.e9-247.e13.	1.5	108
34	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. <i>Neurobiology of Aging</i> , 2009, 30, 1515-1517.	1.5	97
35	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2015, 36, 1605.e7-1605.e12.	1.5	96
36	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	4.5	95

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37	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. <i>Genetics in Medicine</i> , 2017, 19, 45-52.	1.1	94
38	Genetic risk factors for the posterior cortical atrophy variant of Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2016, 12, 862-871.	0.4	93
39	Loss-of-function mutations in <i>RAB39B</i> are associated with typical early-onset Parkinson disease. <i>Neurology: Genetics</i> , 2015, 1, e9.	0.9	90
40	SnapShot: Genetics of Alzheimer's Disease. <i>Cell</i> , 2013, 155, 968-968.e1.	13.5	86
41	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. <i>Neurobiology of Aging</i> , 2014, 35, 2419.e23-2419.e25.	1.5	84
42	Microdeletion in a FAAH pseudogene identified in a patient with high anandamide concentrations and pain insensitivity. <i>British Journal of Anaesthesia</i> , 2019, 123, e249-e253.	1.5	82
43	SnapShot: Genetics of Parkinson's Disease. <i>Cell</i> , 2015, 160, 570-570.e1.	13.5	79
44	Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. <i>Neurobiology of Aging</i> , 2016, 38, 214.e7-214.e10.	1.5	78
45	TYROBP genetic variants in early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 48, 222.e9-222.e15.	1.5	69
46	The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. <i>Current Neurology and Neuroscience Reports</i> , 2018, 18, 67.	2.0	69
47	Alzheimer's disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. <i>Translational Psychiatry</i> , 2019, 9, 154.	2.4	69
48	SnapShot: Genetics of ALS and FTD. <i>Cell</i> , 2015, 160, 798-798.e1.	13.5	68
49	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimer's disease resilience. <i>Genome Medicine</i> , 2017, 9, 100.	3.6	67
50	Frequency of Loss of Function Variants in <i>LRRK2</i> in Parkinson Disease. <i>JAMA Neurology</i> , 2018, 75, 1416.	4.5	66
51	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 460-468.	2.2	66
52	Study protocol: Insight 46 – a neuroscience sub-study of the MRC National Survey of Health and Development. <i>BMC Neurology</i> , 2017, 17, 75.	0.8	64
53	A systematic screening to identify <i>de novo</i> mutations causing sporadic early-onset Parkinson's disease. <i>Human Molecular Genetics</i> , 2015, 24, 6711-6720.	1.4	59
54	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. <i>Human Molecular Genetics</i> , 2014, 23, R47-R53.	1.4	57

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55	Penetrance of Parkinson's Disease in <i>LRRK2</i> p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , 2020, 35, 774-780.	2.2	57
56	Influence of Single Nucleotide Polymorphisms in <i>COMT</i> , <i>MAO-A</i> , and <i>BDNF</i> Genes on Dyskinesias and Levodopa Use in Parkinson's Disease. <i>Neurodegenerative Diseases</i> , 2014, 13, 24-28.	0.8	56
57	Investigating the role of rare coding variability in Mendelian dementia genes (<i>APP</i> , <i>PSEN1</i> , <i>PSEN2</i> , <i>GRN</i>)	1.5	53
58	Analysis of Parkinson disease patients from Portugal for mutations in <i>SNCA</i> , <i>PRKN</i> , <i>PINK1</i> and <i>LRRK2</i> . <i>BMC Neurology</i> , 2008, 8, 1.	0.8	52
59	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. <i>Human Molecular Genetics</i> , 2016, 25, ddd348.	1.4	48
60	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019, 34, 1851-1863.	2.2	47
61	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. <i>Neurobiology of Aging</i> , 2019, 77, 169-177.	1.5	47
62	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome. <i>Nature Communications</i> , 2021, 12, 7342.	5.8	44
63	A rare loss-of-function variant of <i>ADAM17</i> is associated with late-onset familial Alzheimer disease. <i>Molecular Psychiatry</i> , 2020, 25, 629-639.	4.1	42
64	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. <i>JAMA Network Open</i> , 2021, 4, e2030194.	2.8	42
65	Identification of sixteen novel candidate genes for late onset Parkinson's disease. <i>Molecular Neurodegeneration</i> , 2021, 16, 35.	4.4	41
66	<i>ABCA7</i> p.G215S as potential protective factor for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 46, 235.e1-235.e9.	1.5	37
67	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. <i>Alzheimer's and Dementia</i> , 2021, 17, 500-514.	0.4	36
68	Current concepts and controversies in the pathogenesis of Parkinson's disease dementia and Dementia with Lewy Bodies. <i>F1000Research</i> , 2017, 6, 1604.	0.8	35
69	Exome sequencing in a consanguineous family clinically diagnosed with early-onset Alzheimer's disease identifies a homozygous <i>CTSF</i> mutation. <i>Neurobiology of Aging</i> , 2016, 46, 236.e1-236.e6.	1.5	34
70	Influence of Coding Variability in <i>APP</i> -A β Metabolism Genes in Sporadic Alzheimer's Disease. <i>PLoS ONE</i> , 2016, 11, e0150079.	1.1	34
71	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. <i>NeuroImage</i> , 2019, 189, 645-654.	2.1	33
72	A novel human pain insensitivity disorder caused by a point mutation in <i>ZFH2</i> . <i>Brain</i> , 2018, 141, 365-376.	3.7	32

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73	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. <i>Neurobiology of Aging</i> , 2018, 66, 179.e17-179.e29.	1.5	32
74	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2018, 62, 244.e1-244.e8.	1.5	30
75	Heritability and genetic variance of dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2019, 127, 492-501.	2.1	29
76	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021, 90, 35-42.	2.8	29
77	Young-onset parkinsonism due to homozygous duplication of Î±-synuclein in a consanguineous family. <i>Movement Disorders</i> , 2012, 27, 1829-1830.	2.2	27
78	Genetic architecture of common non-Alzheimer's disease dementias. <i>Neurobiology of Disease</i> , 2020, 142, 104946.	2.1	27
79	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. <i>Acta Neuropathologica Communications</i> , 2020, 8, 5.	2.4	27
80	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. <i>Brain</i> , 2022, 145, 1805-1817.	3.7	27
81	Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank. <i>Scientific Reports</i> , 2022, 12, 8404.	1.6	27
82	A nonsense mutation in PRNP associated with clinical Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014, 35, 2656.e13-2656.e16.	1.5	26
83	The Chihuahua dog: A new animal model for neuronal ceroid lipofuscinosis CLN7 disease?. <i>Journal of Neuroscience Research</i> , 2016, 94, 339-347.	1.3	26
84	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. <i>Cortex</i> , 2020, 133, 384-398.	1.1	26
85	Polygenic risk scores for Alzheimer's disease are related to dementia risk in APOE Î¼4 negatives. <i>Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring</i> , 2021, 13, e12142.	1.2	25
86	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. <i>Alzheimer's and Dementia</i> , 2022, 18, 1408-1423.	0.4	24
87	RARS mutations in a sibship with infantile spasms. <i>Epilepsia</i> , 2016, 57, e97-e102.	2.6	23
88	Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. <i>International Journal of Molecular Sciences</i> , 2015, 16, 24629-24655.	1.8	21
89	Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. <i>Neurobiology of Aging</i> , 2017, 49, 215.e1-215.e8.	1.5	21
90	KCNN2 mutation in autosomal dominant tremulous myoclonus-dystonia. <i>European Journal of Neurology</i> , 2020, 27, 1471-1477.	1.7	21

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91	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum τ and pNfH : A Longitudinal Multicentre Study. <i>Annals of Neurology</i> , 2022, 91, 33-47.	2.8	21
92	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. <i>Brain Communications</i> , 2020, 2, .	1.5	20
93	Alzheimer's Disease Genetics: Review of Novel Loci Associated with Disease. <i>Current Genetic Medicine Reports</i> , 2020, 8, 1-16.	1.9	20
94	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. <i>PLoS ONE</i> , 2016, 11, e0162592.	1.1	19
95	Mutation of <i>TBCK</i> causes a rare recessive developmental disorder. <i>Neurology: Genetics</i> , 2016, 2, e76.	0.9	19
96	Multi-infarct dementia of Swedish type is caused by a 3'UTR mutation of COL4A1. <i>Brain</i> , 2017, 140, e29-e29.	3.7	19
97	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. <i>Acta Neuropathologica Communications</i> , 2019, 7, 188.	2.4	19
98	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic <i>C9orf72</i> Repeat Expansion Adult Carriers. <i>Annals of Neurology</i> , 2020, 88, 113-122.	2.8	19
99	Challenge accepted: uncovering the role of rare genetic variants in Alzheimer's disease. <i>Molecular Neurodegeneration</i> , 2022, 17, 3.	4.4	19
100	Assessment of Parkinson's disease risk loci in Greece. <i>Neurobiology of Aging</i> , 2014, 35, 442.e9-442.e16.	1.5	18
101	Clinical, ocular motor, and imaging profile of Niemann-Pick type C heterozygosity. <i>Neurology</i> , 2020, 94, e1702-e1715.	1.5	18
102	A Non- <i>APOE</i> Polygenic Risk Score for Alzheimer's Disease Is Associated With Cerebrospinal Fluid Neurofilament Light in a Representative Sample of Cognitively Unimpaired 70-Year Olds. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2021, 76, 983-990.	1.7	18
103	CLN6 disease caused by the same mutation originating in Pakistan has varying pathology. <i>European Journal of Paediatric Neurology</i> , 2013, 17, 657-660.	0.7	17
104	CYLD variants in frontotemporal dementia associated with severe memory impairment in a Portuguese cohort. <i>Brain</i> , 2020, 143, e67-e67.	3.7	16
105	Genetics of synucleins in neurodegenerative diseases. <i>Acta Neuropathologica</i> , 2021, 141, 471-490.	3.9	16
106	LRP10 in α -synucleinopathies. <i>Lancet Neurology</i> , The, 2018, 17, 1032.	4.9	15
107	Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddon's Syndrome of Unknown Cause. <i>Cerebrovascular Diseases</i> , 2018, 46, 257-264.	0.8	15
108	A comprehensive screening of copy number variability in dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2019, 75, 223.e1-223.e10.	1.5	13

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109	Action Myoclonus and Seizure in Kuforâ€Rakeb Syndrome. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 195-199.	0.8	13
110	A Phenotype of Atypical Apraxia of Speech in a Family Carrying SQSTM1 Mutation. <i>Journal of Alzheimer's Disease</i> , 2014, 43, 625-630.	1.2	12
111	Screening exons 16 and 17 of the amyloid precursor protein gene in sporadic early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 39, 220.e1-220.e7.	1.5	12
112	Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2017, 49, 214.e13-214.e15.	1.5	12
113	An Aged Canid with Behavioral Deficits Exhibits Blood and Cerebrospinal Fluid Amyloid Beta Oligomers. <i>Frontiers in Aging Neuroscience</i> , 2018, 10, 7.	1.7	12
114	LRP10 in α -synucleinopathies. <i>Lancet Neurology</i> , The, 2018, 17, 1032-1033.	4.9	11
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. <i>Journal of Alzheimer's Disease</i> , 2019, 67, 159-167.	1.2	11
116	The clinical syndrome of dystonia with anarthria/aphonia. <i>Parkinsonism and Related Disorders</i> , 2016, 24, 20-27.	1.1	10
117	Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. <i>Neurology: Genetics</i> , 2018, 4, e215.	0.9	10
118	How understudied populations have contributed to our understanding of Alzheimerâ€™s disease genetics. <i>Brain</i> , 2021, 144, 1067-1081.	3.7	10
119	<scp>CLN</scp>8 disease caused by large genomic deletions. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 85-91.	0.6	9
120	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. <i>Molecular Neurodegeneration</i> , 2021, 16, 79.	4.4	9
121	Prion-like α -synuclein pathology in the brain of infants with Krabbe disease. <i>Brain</i> , 2022, 145, 1257-1263.	3.7	9
122	Atypical Parkinsonism-Dystonia Syndrome Caused by a Novel DJ1 Mutation. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 45-49.	0.8	8
123	Disease-related cortical thinning in presymptomatic granulin mutation carriers. <i>NeuroImage: Clinical</i> , 2021, 29, 102540.	1.4	8
124	Genetic variants in glutamate-, $A\beta$, and tau-related pathways determine polygenic risk for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2021, 101, 299.e13-299.e21.	1.5	7
125	Mutations in TYROBP are not a common cause of dementia in a Turkish cohort. <i>Neurobiology of Aging</i> , 2017, 58, 240.e1-240.e3.	1.5	6
126	Genetics of dementia in a Finnish cohort. <i>European Journal of Human Genetics</i> , 2018, 26, 827-837.	1.4	6

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127	<i>AP4S1</i> splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. <i>Neurology: Genetics</i> , 2018, 4, e273.	0.9	6
128	Genotyping of the Alzheimer's Disease Genome-Wide Association Study Index Single Nucleotide Polymorphisms in the Brains for Dementia Research Cohort. <i>Journal of Alzheimer's Disease</i> , 2018, 64, 355-362.	1.2	6
129	Psychiatric Manifestations of <i>ATP13A2</i> Mutations. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 838-841.	0.8	6
130	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. <i>European Journal of Human Genetics</i> , 2021, 29, 663-671.	1.4	6
131	Genetic analysis reveals novel variants for vascular cognitive impairment. <i>Acta Neurologica Scandinavica</i> , 2022, 146, 42-50.	1.0	6
132	Molecular Characterization of Portuguese Patients with Hereditary Cerebellar Ataxia. <i>Cells</i> , 2022, 11, 981.	1.8	6
133	A deletion of IDUA exon 10 in a family of Golden Retriever dogs with an attenuated form of mucopolysaccharidosis type I. <i>Journal of Veterinary Internal Medicine</i> , 2020, 34, 1813-1824.	0.6	4
134	Mutations in a Sibship with Multifocal Polymyoclonus. <i>Tremor and Other Hyperkinetic Movements</i> , 2017, 7, 452.	1.1	4
135	Cognitive composites for genetic frontotemporal dementia: GENFI-Cog. <i>Alzheimer's Research and Therapy</i> , 2022, 14, 10.	3.0	4
136	Genetic Variants and Related Biomarkers in Sporadic Alzheimer's Disease. <i>Current Genetic Medicine Reports</i> , 2015, 3, 19-25.	1.9	3
137	Two pathologically confirmed cases of novel mutations in the MAPT gene causing frontotemporal dementia. <i>Neurobiology of Aging</i> , 2020, 87, 141.e15-141.e20.	1.5	3
138	Patients with progranulin mutations overlap with the progressive dysexecutive syndrome: towards the definition of a frontoparietal dementia phenotype. <i>Brain Communications</i> , 2020, 2, fcaa126.	1.5	3
139	Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. <i>Journal of Clinical Medicine</i> , 2020, 9, 1212.	1.0	3
140	Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. <i>Neuromuscular Disorders</i> , 2021, 31, 891-895.	0.3	2
141	Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort. <i>Cortex</i> , 2022, 150, 12-28.	1.1	2
142	Exome Sequencing of a Portuguese Cohort of Frontotemporal Dementia Patients: Looking Into the ALS-FTD Continuum. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	2
143	Ataxia with oculomotor apraxia is associated with the DNA damage repair pathway. <i>Movement Disorders</i> , 2017, 32, 720-720.	2.2	1
144	Rare variants in TP73 in a frontotemporal dementia cohort link this gene with primary progressive aphasia phenotypes. <i>European Journal of Neurology</i> , 2022, , .	1.7	1

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145	Structural brain splitting is a hallmark of Granulin-related frontotemporal dementia. <i>Neurobiology of Aging</i> , 2022, , .	1.5	1
146	The <sc>CBI&R</sc> detects early behavioural impairment in genetic frontotemporal dementia. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 644-658.	1.7	1
147	P2&158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK. <i>Alzheimer's and Dementia</i> , 2016, 12, P675.	0.4	0
148	P3-091: Investigating SARM1 Variants in Alzheimer&TM's Disease Cohorts. , 2016, 12, P855-P855.		0
149	Mouse models of kufor-rakeb disease link Parkinson's disease closer to neuronal ceroid lipofuscinosis, suggesting lysosomal dysfunction as shared mechanism. <i>Movement Disorders</i> , 2017, 32, 209-209.	2.2	0
150	[P3&101]: MULTI&INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3&TMUTR <i>COL4A1</i> MUTATION. <i>Alzheimer's and Dementia</i> , 2017, 13, P973.	0.4	0
151	[P3&110]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER'S DISEASE. <i>Alzheimer's and Dementia</i> , 2017, 13, P976.	0.4	0
152	[P3&111]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES. <i>Alzheimer's and Dementia</i> , 2017, 13, P977.	0.4	0
153	[P3&112]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER'S DISEASE USING WHOLE&EXOME SEQUENCING. <i>Alzheimer's and Dementia</i> , 2017, 13, P977.	0.4	0
154	[F5&01&02]: GENETICS OF DLB AND RELEVANCE FOR MECHANISMS. <i>Alzheimer's and Dementia</i> , 2017, 13, P1444.	0.4	0
155	[P4&416]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON <i>TYROBP</i>. <i>Alzheimer's and Dementia</i> , 2017, 13, P1490.	0.4	0
156	A comprehensive analysis of copy number variation in a Turkish dementia cohort. <i>Human Genomics</i> , 2021, 15, 48.	1.4	0