

Jose T Bras

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	TREM2 variants in Alzheimer's disease. <i>New England Journal of Medicine</i> , 2013 , 368, 117-27	59.2	1805
153	Diagnosis and management of dementia with Lewy bodies: Fourth consensus report of the DLB Consortium. <i>Neurology</i> , 2017 , 89, 88-100	6.5	1691
152	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. <i>Nature Genetics</i> , 2014 , 46, 989-93	36.3	1261
151	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A β tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019 , 51, 414-430	36.3	917
150	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018 , 360,	33.3	666
149	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, The</i> , 2019 , 18, 1091-1102	24.1	562
148	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. <i>Nature Genetics</i> , 2017 , 49, 1373-1384	36.3	508
147	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. <i>Brain</i> , 2009 , 132, 1783-94	11.2	488
146	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. <i>Nature</i> , 2014 , 505, 550-554	50.4	345
145	A multicenter study of glucocerebrosidase mutations in dementia with Lewy bodies. <i>JAMA Neurology</i> , 2013 , 70, 727-35	17.2	285
144	SNCA variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009 , 65, 610-4	9.4	232
143	Mutation of the parkinsonism gene ATP13A2 causes neuronal ceroid-lipofuscinosis. <i>Human Molecular Genetics</i> , 2012 , 21, 2646-50	5.6	196
142	Mutations in ANO3 cause dominant craniocervical dystonia: ion channel implicated in pathogenesis. <i>American Journal of Human Genetics</i> , 2012 , 91, 1041-50	11	172
141	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. <i>Lancet Neurology, The</i> , 2008 , 7, 207-15	24.1	159
140	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. <i>Human Molecular Genetics</i> , 2014 , 23, 6139-46	5.6	152
139	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2012 , 21, 4996-5009	5.6	145
138	The age factor in Alzheimer's disease. <i>Genome Medicine</i> , 2015 , 7, 106	14.4	137

137	SQSTM1 mutations in French patients with frontotemporal dementia or frontotemporal dementia with amyotrophic lateral sclerosis. <i>JAMA Neurology</i> , 2013 , 70, 1403-10	17.2	131
136	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , 2014 , 137, 2480-92	11.2	127
135	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. <i>Lancet Neurology, The</i> , 2018 , 17, 64-74	24.1	121
134	Insights into TREM2 biology by network analysis of human brain gene expression data. <i>Neurobiology of Aging</i> , 2013 , 34, 2699-714	5.6	119
133	Emerging pathways in genetic Parkinson's disease: Potential role of ceramide metabolism in Lewy body disease. <i>FEBS Journal</i> , 2008 , 275, 5767-73	5.7	109
132	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	11.6	100
131	The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. <i>Lancet Neurology, The</i> , 2018 , 17, 721-730	24.1	92
130	Mutations in PNKP cause recessive ataxia with oculomotor apraxia type 4. <i>American Journal of Human Genetics</i> , 2015 , 96, 474-9	11	90
129	Novel compound heterozygous mutation in TREM2 found in a Turkish frontotemporal dementia-like family. <i>Neurobiology of Aging</i> , 2013 , 34, 2890.e1-5	5.6	90
128	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. <i>Lancet Neurology, The</i> , 2020 , 19, 145-156	24.1	90
127	Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. <i>Nature Reviews Neuroscience</i> , 2012 , 13, 453-64	13.5	89
126	Missense variant in TREML2 protects against Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014 , 35, 1510.e19-2684	5.6	84
125	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. <i>Movement Disorders</i> , 2012 , 27, 526-32	7	80
124	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. <i>Neurobiology of Aging</i> , 2009 , 30, 1515-7	5.6	79
123	SnapShot: genetics of Alzheimer's disease. <i>Cell</i> , 2013 , 155, 968-968.e1	56.2	78
122	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2015 , 36, 1605.e7-12	5.6	70
121	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. <i>Genetics in Medicine</i> , 2017 , 19, 45-52	8.1	68
120	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. <i>Lancet Neurology, The</i> , 2019 , 18, 1103-1111	24.1	68

119	SnapShot: Genetics of Parkinson's disease. <i>Cell</i> , 2015 , 160, 570-570.e1	56.2	67
118	Genetic risk factors for the posterior cortical atrophy variant of Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2016 , 12, 862-71	1.2	64
117	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. <i>Neurology: Genetics</i> , 2015 , 1, e9	3.8	63
116	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. <i>Neurobiology of Aging</i> , 2014 , 35, 2419.e23-2419.e25	5.6	63
115	Genetic analysis of inherited leukodystrophies: genotype-phenotype correlations in the CSF1R gene. <i>JAMA Neurology</i> , 2013 , 70, 875-882	17.2	58
114	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	5.6	54
113	SnapShot: Genetics of ALS and FTD. <i>Cell</i> , 2015 , 160, 798-798.e1	56.2	54
112	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	5.4	51
111	Expanding Parkinson's disease genetics: novel risk loci, genomic context, causal insights and heritable risk		51
110	TYROBP genetic variants in early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016 , 48, 222.e9-222.e15	5.6	51
109	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. <i>JAMA Neurology</i> , 2018 , 75, 1416-1422	14.2	50
108	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	5.6	49
107	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. <i>Human Molecular Genetics</i> , 2014 , 23, R47-53	5.6	48
106	Investigating the role of rare coding variability in Mendelian dementia genes (APP, PSEN1, PSEN2, GRN, MAPT, and PRNP) in late-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014 , 35, 2881.e1-2881.e6	5.6	45
105	Study protocol: Insight 46 - a neuroscience sub-study of the MRC National Survey of Health and Development. <i>BMC Neurology</i> , 2017 , 17, 75	3.1	42
104	The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. <i>Current Neurology and Neuroscience Reports</i> , 2018 , 18, 67	6.6	41
103	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019 , 34, 460-468	7	40
102	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimer's disease resilience. <i>Genome Medicine</i> , 2017 , 9, 100	14.4	40

101	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, 5483-5489	5.6	40
100	Influence of single nucleotide polymorphisms in COMT, MAO-A and BDNF genes on dyskinesias and levodopa use in Parkinson's disease. <i>Neurodegenerative Diseases</i> , 2014 , 13, 24-8	2.3	40
99	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016 , 46, 235.e1-9	5.6	33
98	Analysis of Parkinson disease patients from Portugal for mutations in SNCA, PRKN, PINK1 and LRRK2. <i>BMC Neurology</i> , 2008 , 8, 1	3.1	32
97	Alzheimer's disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. <i>Translational Psychiatry</i> , 2019 , 9, 154	8.6	31
96	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , 2020 , 35, 774-780	7	27
95	A rare loss-of-function variant of ADAM17 is associated with late-onset familial Alzheimer disease. <i>Molecular Psychiatry</i> , 2020 , 25, 629-639	15.1	27
94	A systematic screening to identify de novo mutations causing sporadic early-onset Parkinson's disease. <i>Human Molecular Genetics</i> , 2015 , 24, 6711-20	5.6	26
93	Influence of Coding Variability in APP- β Metabolism Genes in Sporadic Alzheimer's Disease. <i>PLoS ONE</i> , 2016 , 11, e0150079	3.7	26
92	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2018 , 62, 244.e1-244.e8	5.6	25
91	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. <i>Neurobiology of Aging</i> , 2019 , 77, 169-177	5.6	24
90	Current concepts and controversies in the pathogenesis of Parkinson's disease dementia and Dementia with Lewy Bodies. <i>F1000Research</i> , 2017 , 6, 1604	3.6	23
89	A novel human pain insensitivity disorder caused by a point mutation in ZFH2. <i>Brain</i> , 2018 , 141, 365-376	11.2	23
88	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	5.6	23
87	Tremor-ataxia with central hypomyelination (TACH): dystonia as a new clinical feature. <i>Movement Disorders</i> , 2012 , 27, 1829-30	7	23
86	Nonsense mutation in PRNP associated with clinical Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014 , 35, 2656.e13-2656.e16	5.6	22
85	The Chihuahua dog: A new animal model for neuronal ceroid lipofuscinosis CLN7 disease?. <i>Journal of Neuroscience Research</i> , 2016 , 94, 339-47	4.4	22
84	Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. <i>International Journal of Molecular Sciences</i> , 2015 , 16, 24629-55	6.3	19

83	Exome sequencing in a consanguineous family clinically diagnosed with early-onset Alzheimer's disease identifies a homozygous CTSF mutation. <i>Neurobiology of Aging</i> , 2016 , 46, 236.e1-6	5.6	19
82	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	7.63	18
81	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. <i>NeuroImage</i> , 2019 , 189, 645-654	7.9	18
80	RARS2 mutations in a sibship with infantile spasms. <i>Epilepsia</i> , 2016 , 57, e97-e102	6.4	18
79	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021 , 78, 464-472	17.2	17
78	Genetic architecture of common non-Alzheimer's disease dementias. <i>Neurobiology of Disease</i> , 2020 , 142, 104946	7.5	16
77	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	3.7	16
76	Heritability and genetic variance of dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2019 , 127, 492-501	5.91	15
75	Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. <i>Neurobiology of Aging</i> , 2017 , 49, 215.e1-215.e8	5.6	15
74	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. <i>Acta Neuropathologica Communications</i> , 2020 , 8, 5	7.3	15
73	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. <i>JAMA Network Open</i> , 2021 , 4, e2030194	10.4	14
72	LRP10 in Synucleinopathies. <i>Lancet Neurology</i> , 2018 , 17, 1032	24.1	14
71	Mutation of TBCK causes a rare recessive developmental disorder. <i>Neurology: Genetics</i> , 2016 , 2, e76	3.8	13
70	Multi-infarct dementia of Swedish type is caused by a 3'UTR mutation of COL4A1. <i>Brain</i> , 2017 , 140, e29	11.2	12
69	Clinical, ocular motor, and imaging profile of Niemann-Pick type C heterozygosity. <i>Neurology</i> , 2020 , 94, e1702-e1715	6.5	12
68	Alzheimer's Disease Genetics: Review of Novel Loci Associated with Disease. <i>Current Genetic Medicine Reports</i> , 2020 , 8, 1-16	2.2	12
67	Assessment of Parkinson's disease risk loci in Greece. <i>Neurobiology of Aging</i> , 2014 , 35, 442.e9-442.e16	5.6	12
66	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. <i>Annals of Neurology</i> , 2020 , 88, 113-122	9.4	11

65	Action Myoclonus and Seizure in Kufor-Rakeb Syndrome. <i>Movement Disorders Clinical Practice</i> , 2018 , 5, 195-199	2.2	11
64	KCNN2 mutation in autosomal-dominant tremulous myoclonus-dystonia. <i>European Journal of Neurology</i> , 2020 , 27, 1471-1477	6	10
63	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	5.6	10
62	A phenotype of atypical apraxia of speech in a family carrying SQSTM1 mutation. <i>Journal of Alzheimer's Disease</i> , 2015 , 43, 625-30	4.3	10
61	A comprehensive screening of copy number variability in dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2019 , 75, 223.e1-223.e10	5.6	10
60	Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddon's Syndrome of Unknown Cause. <i>Cerebrovascular Diseases</i> , 2018 , 46, 257-264	3.2	10
59	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.e1-7	5.6	9
58	LRP10 in Synucleinopathies. <i>Lancet Neurology</i> , 2018 , 17, 1032-1033	24.1	9
57	Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. <i>Neurology: Genetics</i> , 2018 , 4, e215	3.8	8
56	CLN6 disease caused by the same mutation originating in Pakistan has varying pathology. <i>European Journal of Paediatric Neurology</i> , 2013 , 17, 657-60	3.8	8
55	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. <i>Alzheimer's and Dementia</i> , 2021 , 17, 500-514	1.2	8
54	The clinical syndrome of dystonia with anarthria/aphonia. <i>Parkinsonism and Related Disorders</i> , 2016 , 24, 20-7	3.6	7
53	An Aged Canid with Behavioral Deficits Exhibits Blood and Cerebrospinal Fluid Amyloid Beta Oligomers. <i>Frontiers in Aging Neuroscience</i> , 2018 , 10, 7	5.3	7
52	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. <i>Cortex</i> , 2020 , 133, 384-398	3.8	7
51	A comprehensive assessment of benign genetic variability for neurodegenerative disorders		7
50	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	4.3	7
49	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	4.3	6
48	Atypical Parkinsonism-Dystonia Syndrome Caused by a Novel DJ1 Mutation. <i>Movement Disorders Clinical Practice</i> , 2014 , 1, 45-49	2.2	6

47	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. <i>Brain Communications</i> , 2020 , 2,	4.5	6
46	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021 , 90, 35-42	9.4	6
45	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 188	7.3	6
44	Genetics of synucleins in neurodegenerative diseases. <i>Acta Neuropathologica</i> , 2021 , 141, 471-490	14.3	6
43	Genetics of dementia in a Finnish cohort. <i>European Journal of Human Genetics</i> , 2018 , 26, 827-837	5.3	5
42	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	5.6	5
41	CYLD variants in frontotemporal dementia associated with severe memory impairment in a Portuguese cohort. <i>Brain</i> , 2020 , 143, e67	11.2	5
40	Polygenic risk scores for Alzheimer's disease are related to dementia risk in APOE e4 negatives. <i>Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring</i> , 2021 , 13, e12142	5.2	5
39	CLN8 disease caused by large genomic deletions. <i>Molecular Genetics & Genomic Medicine</i> , 2017 , 5, 85-91	2.3	4
38	Mutations in a Sibship with Multifocal Polymyoclonus. <i>Tremor and Other Hyperkinetic Movements</i> , 2017 , 7, 452	2	4
37	Psychiatric Manifestations of Mutations. <i>Movement Disorders Clinical Practice</i> , 2020 , 7, 838-841	2.2	4
36	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. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2021 , 76, 983-990	6.4	4
35	splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. <i>Neurology: Genetics</i> , 2018 , 4, e273	3.8	4
34	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. <i>Brain</i> , 2021 ,	11.2	3
33	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	4.5	3
32	Identification of sixteen novel candidate genes for late onset Parkinson's disease. <i>Molecular Neurodegeneration</i> , 2021 , 16, 35	19	3
31	Genetic variants in glutamate-, A β and tau-related pathways determine polygenic risk for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2021 , 101, 299.e13-299.e21	5.6	3
30	Prion-like β synuclein pathology in the brain of infants with Krabbe disease.. <i>Brain</i> , 2022 ,	11.2	2

29	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. <i>Alzheimer's and Dementia</i> , 2021 ,	1.2	2
28	Stratifying the Presymptomatic Phase of Genetic Frontotemporal Dementia by Serum NFL and pNFH: A Longitudinal Multicentre Study. <i>Annals of Neurology</i> , 2021 ,	9.4	2
27	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	5.6	2
26	How understudied populations have contributed to our understanding of Alzheimer's disease genetics. <i>Brain</i> , 2021 , 144, 1067-1081	11.2	2
25	Disease-related cortical thinning in presymptomatic granulin mutation carriers. <i>NeuroImage: Clinical</i> , 2021 , 29, 102540	5.3	2
24	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. <i>Nature Communications</i> , 2021 , 12, 7342	17.4	2
23	Ataxia with oculomotor apraxia is associated with the DNA damage repair pathway. <i>Movement Disorders</i> , 2017 , 32, 720	7	1
22	Genetic Variants and Related Biomarkers in Sporadic Alzheimer's Disease. <i>Current Genetic Medicine Reports</i> , 2015 , 3, 19-25	2.2	1
21	Novel Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	1
20	Challenge accepted: uncovering the role of rare genetic variants in Alzheimer's disease.. <i>Molecular Neurodegeneration</i> , 2022 , 17, 3	19	1
19	How understudied populations have contributed to our understanding of Alzheimer's disease genetics		1
18	Exome sequencing identifies rare damaging variants in the ATP8B4 and ABCA1 genes as novel risk factors for Alzheimer's Disease		1
17	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	3.1	1
16	Cognitive composites for genetic frontotemporal dementia: GENFI-Cog.. <i>Alzheimer's Research and Therapy</i> , 2022 , 14, 10	9	0
15	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. <i>Molecular Neurodegeneration</i> , 2021 , 16, 79	19	0
14	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. <i>European Journal of Human Genetics</i> , 2021 , 29, 663-671	5.3	0
13	Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank.. <i>Scientific Reports</i> , 2022 , 12, 8404	4.9	0
12	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	7	

- 11 [P3001]: MULTI-INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3'UTR COL4A1 MUTATION **2017**, 13, P973-P973
- 10 [P3010]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER'S DISEASE **2017**, 13, P976-P977
- 9 [P3011]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES **2017**, 13, P977-P977
- 8 [P3012]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER'S DISEASE USING WHOLE-EXOME SEQUENCING **2017**, 13, P977-P977
- 7 [F50102]: GENETICS OF DLB AND RELEVANCE FOR MECHANISMS **2017**, 13, P1444
- 6 [P4016]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON TYROBP **2017**, 13, P1490-P1491
- 5 Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort.. *Cortex*, **2022**, 150, 12-28 3.8
- 4 Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. *Neuromuscular Disorders*, **2021**, 31, 891-895 2.9
- 3 P2-158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK **2016**, 12, P675-P675
- 2 P3-091: Investigating SARM1 Variants in Alzheimer's Disease Cohorts **2016**, 12, P855-P855
- 1 A comprehensive analysis of copy number variation in a Turkish dementia cohort. *Human Genomics*, **2021**, 15, 48 6.8