Jose T Bras

List of Publications by Year
in descending order

156 papers	20,872 citations	31976 53 h-index	11939 134 g-index
176 all docs	176 docs citations	176 times ranked	25390 citing authors

#	Article	IF	Citations
1	A data-driven disease progression model of fluid biomarkers in genetic frontotemporal dementia. Brain, 2022, 145, 1805-1817.	7.6	27
2	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.	5.3	21
3	Challenge accepted: uncovering the role of rare genetic variants in Alzheimerâ€™s disease. Molecular Neurodegeneration, 2022, 17, 3.	10.8	19
4	Cognitive composites for genetic frontotemporal dementia: GENFI-Cog. Alzheimer's Research and Therapy, 2022, 14, 10.	6.2	4
5	Rare variants in TP73 in a frontotemporal dementia cohort link this gene with primary progressive aphasia phenotypes. European Journal of Neurology, 2022, , .	3.3	1
6	Prion-like Î±-synuclein pathology in the brain of infants with Krabbe disease. Brain, 2022, 145, 1257-1263.	7.6	9
7	Examining empathy deficits across familial forms of frontotemporal dementia within the GENFI cohort. Cortex, 2022, 150, 12-28.	2.4	2
8	Conceptual framework for the definition of preclinical and prodromal frontotemporal dementia. Alzheimer's and Dementia, 2022, 18, 1408-1423.	0.8	24
9	Structural brain splitting is a hallmark of Granulin-related frontotemporal dementia. Neurobiology of Aging, 2022, , .	3.1	1
10	Genetic analysis reveals novel variants for vascular cognitive impairment. Acta Neurologica Scandinavica, 2022, 146, 42-50.	2.1	6
11	The <scp>CBIâ€R</scp> detects early behavioural impairment in genetic frontotemporal dementia. Annals of Clinical and Translational Neurology, 2022, 9, 644-658.	3.7	1
12	Molecular Characterization of Portuguese Patients with Hereditary Cerebellar Ataxia. Cells, 2022, 11, 981.	4.1	6
13	Genome-wide association of polygenic risk extremes for Alzheimer's disease in the UK Biobank. Scientific Reports, 2022, 12, 8404.	3.3	27
14	Genetics of synucleins in neurodegenerative diseases. Acta Neuropathologica, 2021, 141, 471-490.	7.7	16
15	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. European Journal of Human Genetics, 2021, 29, 663-671.	2.8	6
16	Brain functional network integrity sustains cognitive function despite atrophy in presymptomatic genetic frontotemporal dementia. Alzheimer's and Dementia, 2021, 17, 500-514.	0.8	36
17	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.	3.1	7
18	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.	3.6	18

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19	Progression of Behavioral Disturbances and Neuropsychiatric Symptoms in Patients With Genetic Frontotemporal Dementia. JAMA Network Open, 2021, 4, e2030194.	5.9	42
20	How understudied populations have contributed to our understanding of Alzheimerâ€™s disease genetics. Brain, 2021, 144, 1067-1081.	7.6	10
21	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	9.0	95
22	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	5.3	29
23	Vasculitic peripheral neuropathy in deficiency of adenosine deaminase 2. Neuromuscular Disorders, 2021, 31, 891-895.	0.6	2
24	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	10.8	41
25	A comprehensive analysis of copy number variation in a Turkish dementia cohort. Human Genomics, 2021, 15, 48.	2.9	0
26	Disease-related cortical thinning in presymptomatic granulin mutation carriers. NeuroImage: Clinical, 2021, 29, 102540.	2.7	8
27	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.	2.4	25
28	A panel of CSF proteins separates genetic frontotemporal dementia from presymptomatic mutation carriers: a GENFI study. Molecular Neurodegeneration, 2021, 16, 79.	10.8	9
29	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44
30	A rare loss-of-function variant of ADAM17 is associated with late-onset familial Alzheimer disease. Molecular Psychiatry, 2020, 25, 629-639.	7.9	42
31	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.	10.2	175
32	Two pathologically confirmed cases of novel mutations in the MAPT gene causing frontotemporal dementia. Neurobiology of Aging, 2020, 87, 141.e15-141.e20.	3.1	3
33	CYLD variants in frontotemporal dementia associated with severe memory impairment in a Portuguese cohort. Brain, 2020, 143, e67-e67.	7.6	16
34	Patients with progranulin mutations overlap with the progressive dysexecutive syndrome: towards the definition of a frontoparietal dementia phenotype. Brain Communications, 2020, 2, fcaa126.	3.3	3
35	Psychiatric Manifestations of <scp>ATP13A2</scp> Mutations. Movement Disorders Clinical Practice, 2020, 7, 838-841.	1.5	6
36	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.	1.6	4

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37	Analysis of brain atrophy and local gene expression in genetic frontotemporal dementia. Brain Communications, 2020, 2, .	3.3	20
38	Clinical, ocular motor, and imaging profile of Niemann-Pick type C heterozygosity. Neurology, 2020, 94, e1702-e1715.	1.1	18
39	KCNN2 mutation in autosomalâ€dominant tremulous myoclonusâ€dystonia. European Journal of Neurology, 2020, 27, 1471-1477.	3.3	21
40	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	3.9	57
41	Alzheimerâ€™s Disease Genetics: Review of Novel Loci Associated with Disease. Current Genetic Medicine Reports, 2020, 8, 1-16.	1.9	20
42	Novel MAG Variant Causes Cerebellar Ataxia with Oculomotor Apraxia: Molecular Basis and Expanded Clinical Phenotype. Journal of Clinical Medicine, 2020, 9, 1212.	2.4	3
43	Faster Cortical Thinning and Surface Area Loss in Presymptomatic and Symptomatic C9orf72 Repeat Expansion Adult Carriers. Annals of Neurology, 2020, 88, 113-122.	5.3	19
44	Social cognition impairment in genetic frontotemporal dementia within the GENFI cohort. Cortex, 2020, 133, 384-398.	2.4	26
45	Genetic architecture of common non-Alzheimerâ€™s disease dementias. Neurobiology of Disease, 2020, 142, 104946.	4.4	27
46	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. Acta Neuropathologica Communications, 2020, 8, 5.	5.2	27
47	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.	10.2	1,414
48	Serum neurofilament light chain in genetic frontotemporal dementia: a longitudinal, multicentre cohort study. Lancet Neurology, The, 2019, 18, 1103-1111.	10.2	128
49	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.	3.9	47
50	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	3.9	66
51	The inner fluctuations of the brain in presymptomatic Frontotemporal Dementia: The chronnectome fingerprint. NeuroImage, 2019, 189, 645-654.	4.2	33
52	Alzheimerâ€™s disease polygenic risk score as a predictor of conversion from mild-cognitive impairment. Translational Psychiatry, 2019, 9, 154.	4.8	69
53	Microdeletion in a FAAH pseudogene identified in a patient with high anandamide concentrations and pain insensitivity. British Journal of Anaesthesia, 2019, 123, e249-e253.	3.4	82
54	Heritability and genetic variance of dementia with Lewy bodies. Neurobiology of Disease, 2019, 127, 492-501.	4.4	29

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55	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.	21.4	1,962
56	An AARS variant as the likely cause of Swedish type hereditary diffuse leukoencephalopathy with spheroids. Acta Neuropathologica Communications, 2019, 7, 188.	5.2	19
57	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.	2.6	11
58	Functional network resilience to pathology in presymptomatic genetic frontotemporal dementia. Neurobiology of Aging, 2019, 77, 169-177.	3.1	47
59	A comprehensive screening of copy number variability in dementia with Lewy bodies. Neurobiology of Aging, 2019, 75, 223.e1-223.e10.	3.1	13
60	Genetics of dementia in a Finnish cohort. European Journal of Human Genetics, 2018, 26, 827-837.	2.8	6
61	Late-onset and acute presentation of Brown-Vialetto-Van Laere syndrome in a Brazilian family. Neurology: Genetics, 2018, 4, e215.	1.9	10
62	A novel human pain insensitivity disorder caused by a point mutation in ZFHX2. Brain, 2018, 141, 365-376.	7.6	32
63	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. Neurobiology of Aging, 2018, 66, 179.e17-179.e29.	3.1	32
64	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. Neurobiology of Aging, 2018, 62, 244.e1-244.e8.	3.1	30
65	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. Lancet Neurology, The, 2018, 17, 64-74.	10.2	195
66	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	10.2	15
67	Adenosine Deaminase Two and Immunoglobulin M Accurately Differentiate Adult Sneddonâ€™s Syndrome of Unknown Cause. Cerebrovascular Diseases, 2018, 46, 257-264.	1.7	15
68	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032-1033.	10.2	11
69	AP4S1 splice-site mutation in a case of spastic paraplegia type 52 with polymicrogyria. Neurology: Genetics, 2018, 4, e273.	1.9	6
70	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.	2.6	6
71	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	9.0	66
72	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085

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73	An Aged Canid with Behavioral Deficits Exhibits Blood and Cerebrospinal Fluid Amyloid Beta Oligomers. Frontiers in Aging Neuroscience, 2018, 10, 7.	3.4	12
74	The role of TREM2 in Alzheimer's disease and other neurodegenerative disorders. Lancet Neurology, The, 2018, 17, 721-730.	10.2	161
75	The Genetics of Dementia with Lewy Bodies: Current Understanding and Future Directions. Current Neurology and Neuroscience Reports, 2018, 18, 67.	4.2	69
76	Action Myoclonus and Seizure in Kuforâ€Rakeb Syndrome. Movement Disorders Clinical Practice, 2018, 5, 195-199.	1.5	13
77	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
78	<scp>CLN</scp>8 disease caused by large genomic deletions. Molecular Genetics & Genomic Medicine, 2017, 5, 85-91.	1.2	9
79	Mouse models of kufor-rakeb disease link Parkinson's disease closer to neuronal ceroid lipofuscinosis, suggesting lysosomal dysfunction as shared mechanism. Movement Disorders, 2017, 32, 209-209.	3.9	0
80	Ataxia with oculomotor apraxia is associated with the DNA damage repair pathway. Movement Disorders, 2017, 32, 720-720.	3.9	1
81	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.	3.1	108
82	Diagnosis and management of dementia with Lewy bodies. Neurology, 2017, 89, 88-100.	1.1	2,805
83	Multi-infarct dementia of Swedish type is caused by a 3â€™UTR mutation of COL4A1. Brain, 2017, 140, e29-e29.	7.6	19
84	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	21.4	783
85	Mutations in TYROBP are not a common cause of dementia in a Turkish cohort. Neurobiology of Aging, 2017, 58, 240.e1-240.e3.	3.1	6
86	[P3â€“101]: MULTIâ€INFARCT DEMENTIA OF SWEDISH TYPE IS CAUSED BY 3â€™UTR COL4A1 MUTATION. Alzheimer's and Dementia, 2017, 13, P973.	0.8	0
87	Study protocol: Insight 46 â€“ a neuroscience sub-study of the MRC National Survey of Health and Development. BMC Neurology, 2017, 17, 75.	1.8	64
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.	3.1	12
89	Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. Neurobiology of Aging, 2017, 49, 215.e1-215.e8.	3.1	21
90	[P3â€“110]: CALCULATING POLYGENIC RISK FOR INDIVIDUALS WITH SPORADIC EARLY ONSET ALZHEIMER's DISEASE. Alzheimer's and Dementia, 2017, 13, P976.	0.8	0

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91	[P3â€“111]: NOVEL CANDIDATE GENES FOR DEMENTIA WITH LEWY BODIES. Alzheimer's and Dementia, 2017, 13, P977.	0.8	0
92	[P3â€“112]: INVESTIGATING GENETIC VARIATION IN ALZHEIMER's DISEASE USING WHOLEâ€EXOME SEQUENCING. Alzheimer's and Dementia, 2017, 13, P977.	0.8	0
93	[F5â€“01â€“02]: GENETICS OF DLB AND RELEVANCE FOR MECHANISMS. Alzheimer's and Dementia, 2017, 13, P1444.	0.8	0
94	[P4â€“416]: GENETIC CHARACTERIZATION OF A TURKISH DEMENTIA COHORT: FOCUS ON TYROBP. Alzheimer's and Dementia, 2017, 13, P1490.	0.8	0
95	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimerâ€™s disease resilience. Genome Medicine, 2017, 9, 100.	8.2	67
96	Current concepts and controversies in the pathogenesis of Parkinsonâ€™s disease dementia and Dementia with Lewy Bodies. F1000Research, 2017, 6, 1604.	1.6	35
97	Mutations in a Sibship with Multifocal Polymyoclonus. Tremor and Other Hyperkinetic Movements, 2017, 7, 452.	2.0	4
98	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. PLoS ONE, 2016, 11, e0162592.	2.5	19
99	The Chihuahua dog: A new animal model for neuronal ceroid lipofuscinosis CLN7 disease?. Journal of Neuroscience Research, 2016, 94, 339-347.	2.9	26
100	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.	3.1	34
101	Mutation of TBCK causes a rare recessive developmental disorder. Neurology: Genetics, 2016, 2, e76.	1.9	19
102	P2â€•158: LINKAGE AND WHOLE GENOME SEQUENCE ANALYSIS OF ALZHEIMER'S DISEASE RESILIENCE AND RISK. Alzheimer's and Dementia, 2016, 12, P675.	0.8	0
103	P3-091: Investigating SARM1 Variants in Alzheimerâ€™s Disease Cohorts. , 2016, 12, P855-P855.		0
104	Genetic risk factors for the posterior cortical atrophy variant of Alzheimer's disease. Alzheimer's and Dementia, 2016, 12, 862-871.	0.8	93
105	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	3.1	69
106	<scp>RARS</scp>2 mutations in a sibship with infantile spasms. Epilepsia, 2016, 57, e97-e102.	5.1	23
107	Additional rare variant analysis in Parkinsonâ€™s disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. Human Molecular Genetics, 2016, 25, ddw348.	2.9	48
108	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. Neurobiology of Aging, 2016, 46, 235.e1-235.e9.	3.1	37

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109	The clinical syndrome of dystonia with anarthria/aphonia. Parkinsonism and Related Disorders, 2016, 24, 20-27.	2.2	10
110	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.	3.1	78
111	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.	3.1	12
112	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. PLoS Medicine, 2016, 13, e1001976.	8.4	150
113	Influence of Coding Variability in APP-AÎ² Metabolism Genes in Sporadic Alzheimerâ€™s Disease. PLoS ONE, 2016, 11, e0150079.	2.5	34
114	The age factor in Alzheimerâ€™s disease. Genome Medicine, 2015, 7, 106.	8.2	271
115	Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. International Journal of Molecular Sciences, 2015, 16, 24629-24655.	4.1	21
116	SnapShot: Genetics of ALS and FTD. Cell, 2015, 160, 798-798.e1.	28.9	68
117	SnapShot: Genetics of Parkinsonâ€™s Disease. Cell, 2015, 160, 570-570.e1.	28.9	79
118	Mutations in PNKP Cause Recessive Ataxia with Oculomotor Apraxia Type 4. American Journal of Human Genetics, 2015, 96, 474-479.	6.2	127
119	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. Neurology: Genetics, 2015, 1, e9.	1.9	90
120	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	3.1	96
121	Genetic Variants and Related Biomarkers in Sporadic Alzheimerâ€™s Disease. Current Genetic Medicine Reports, 2015, 3, 19-25.	1.9	3
122	A systematic screening to identifyde novomutations causing sporadic early-onset Parkinson's disease. Human Molecular Genetics, 2015, 24, 6711-6720.	2.9	59
123	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. Neurodegenerative Diseases, 2014, 13, 24-28.	1.4	56
124	A Phenotype of Atypical Apraxia of Speech in a Family Carrying SQSTM1 Mutation. Journal of Alzheimer's Disease, 2014, 43, 625-630.	2.6	12
125	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. Human Molecular Genetics, 2014, 23, 6139-6146.	2.9	178
126	Atypical Parkinsonism-Dystonia Syndrome Caused by a Novel DJ1 Mutation. Movement Disorders Clinical Practice, 2014, 1, 45-49.	1.5	8

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127	Investigating the role of rare coding variability in Mendelian dementia genes (APP , PSEN1 , PSEN2 , GRN) Tj ETQq1 1 0.784314 rgBT	3.1	53
128	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. Human Molecular Genetics, 2014, 23, R47-R53.	2.9	57
129	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
130	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993.	21.4	1,685
131	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
132	Assessment of Parkinson's disease risk loci in Greece. Neurobiology of Aging, 2014, 35, 442.e9-442.e16.	3.1	18
133	Homozygous TREM2 mutation in a family with atypical frontotemporal dementia. Neurobiology of Aging, 2014, 35, 2419.e23-2419.e25.	3.1	84
134	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
135	A nonsense mutation in PRNP associated with clinical Alzheimer's disease. Neurobiology of Aging, 2014, 35, 2656.e13-2656.e16.	3.1	26
136	SnapShot: Genetics of Alzheimerâ€™s Disease. Cell, 2013, 155, 968-968.e1.	28.9	86
137	Insights into TREM2 biology by network analysis of human brain gene expression data. Neurobiology of Aging, 2013, 34, 2699-2714.	3.1	145
138	TREM2 Variants in Alzheimer's Disease. New England Journal of Medicine, 2013, 368, 117-127.	27.0	2,385
139	CLN6 disease caused by the same mutation originating in Pakistan has varying pathology. European Journal of Paediatric Neurology, 2013, 17, 657-660.	1.6	17
140	A novel compound heterozygous mutation in TREM2 found in a Turkish frontotemporal dementia-like family. Neurobiology of Aging, 2013, 34, 2890.e1-2890.e5.	3.1	113
141	Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875.	9.0	147
142	SQSTM1Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. JAMA Neurology, 2013, 70, 1403-10.	9.0	153
143	A Multicenter Study of Glucocerebrosidase Mutations in Dementia With Lewy Bodies. JAMA Neurology, 2013, 70, 727.	9.0	374
144	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	2.9	176

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145	Youngâ€onset parkinsonism due to homozygous duplication of Î±â€synuclein in a consanguineous family. Movement Disorders, 2012, 27, 1829-1830.	3.9	27
146	Mutation of the parkinsonism gene ATP13A2 causes neuronal ceroid-lipofuscinosis. Human Molecular Genetics, 2012, 21, 2646-2650.	2.9	231
147	Use of next-generation sequencing and other whole-genome strategies to dissect neurological disease. Nature Reviews Neuroscience, 2012, 13, 453-464.	10.2	110
148	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050.	6.2	224
149	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532.	3.9	108
150	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. Brain, 2009, 132, 1783-1794.	7.6	612
151	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	5.3	257
152	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. Neurobiology of Aging, 2009, 30, 1515-1517.	3.1	97
153	Emerging pathways in genetic Parkinson's disease: Potential role of ceramide metabolism in Lewy body disease. FEBS Journal, 2008, 275, 5767-5773.	4.7	121
154	Analysis of Parkinson disease patients from Portugal for mutations in SNCA, PRKN, PINK1 and LRRK2. BMC Neurology, 2008, 8, 1.	1.8	52
155	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.	10.2	202
156	Exome Sequencing of a Portuguese Cohort of Frontotemporal Dementia Patients: Looking Into the ALS-FTD Continuum. Frontiers in Neurology, 0, 13, .	2.4	2