Nabec North American Brain Expressio

List of Publications by Year
in descending order

136 papers	26,348 citations	16451 64 h-index	12272 133 g-index
157 all docs	157 docs citations	157 times ranked	32823 citing authors

#	ARTICLE	IF	CITATIONS
1	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. Brain, 2022, 145, 1757-1762.	7.6	17
2	Heterozygous PRKN mutations are common but do not increase the risk of Parkinsonâ€™s disease. Brain, 2022, 145, 2077-2091.	7.6	26
3	ATXN2 intermediate expansions in amyotrophic lateral sclerosis. Brain, 2022, 145, 2671-2676.	7.6	16
4	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
5	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. Lancet Neurology, The, 2021, 20, 107-116.	10.2	62
6	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. Neurology, 2021, 96, e600-e609.	1.1	23
7	Replication assessment of NUS1 variants in Parkinson's disease. Neurobiology of Aging, 2021, 101, 300.e1-300.e3.	3.1	3
8	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459.	3.9	16
9	The Parkinson's Disease <scp>DNA</scp> Variant Browser. Movement Disorders, 2021, 36, 1250-1258.	3.9	11
10	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. Nature Genetics, 2021, 53, 294-303.	21.4	198
11	Exploring dementia and neuronal ceroid lipofuscinosis genes in 100 FTD-like patients from 6 towns and rural villages on the Adriatic Sea cost of Apulia. Scientific Reports, 2021, 11, 6353.	3.3	7
12	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
13	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	5.3	29
14	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. Movement Disorders, 2021, 36, 1795-1804.	3.9	60
15	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	10.8	41
16	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
17	RNA sequencing of whole blood reveals early alterations in immune cells and gene expression in Parkinsonâ€™s disease. Nature Aging, 2021, 1, 734-747.	11.6	18
18	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44

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19	Genetic modifiers of risk and age at onset in GBA associated Parkinsonâ€™s disease and Lewy body dementia. Brain, 2020, 143, 234-248.	7.6	149
20	A SINE-VNTR-Alu in the LRIG2 Promoter Is Associated with Gene Expression at the Locus. International Journal of Molecular Sciences, 2020, 21, 8486.	4.1	6
21	The Parkinson's Disease <scp>Genomeâ€Wide</scp> Association Study Locus Browser. Movement Disorders, 2020, 35, 2056-2067.	3.9	68
22	Large-scale pathway specific polygenic risk and transcriptomic community network analysis identifies novel functional pathways in Parkinson disease. Acta Neuropathologica, 2020, 140, 341-358.	7.7	68
23	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. Nature Communications, 2020, 11, 1041.	12.8	22
24	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
25	MIDN locus structural variants and Parkinson's Disease risk. Annals of Clinical and Translational Neurology, 2020, 7, 602-603.	3.7	5
26	Genomewide association study of Parkinson's disease clinical biomarkers in 12 longitudinal patients' cohorts. Movement Disorders, 2019, 34, 1839-1850.	3.9	122
27	The Parkinson's Disease Mendelian Randomization Research Portal. Movement Disorders, 2019, 34, 1864-1872.	3.9	50
28	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
29	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
30	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
31	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. Movement Disorders, 2019, 34, 1333-1344.	3.9	21
32	Mitochondria function associated genes contribute to Parkinsonâ€™s Disease risk and later age at onset. Npj Parkinson's Disease, 2019, 5, 8.	5.3	95
33	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinsonâ€™s disease heritability. Npj Parkinson's Disease, 2019, 5, 6.	5.3	83
34	Parkinson's disease age at onset genomeâ€wide association study: Defining heritability, genetic loci, and Î±â€synuclein mechanisms. Movement Disorders, 2019, 34, 866-875.	3.9	258
35	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
36	Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. JAMA Neurology, 2018, 75, 591.	9.0	93

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37	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	3.1	30
38	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
39	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
40	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	9.0	66
41	A comprehensive analysis of SNCAâ€related genetic risk in sporadic parkinson disease. Annals of Neurology, 2018, 84, 117-129.	5.3	50
42	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	12.8	250
43	Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22.	8.8	96
44	Genetics of early-onset Parkinson's disease in Finland: exome sequencing and genome-wide association study. Neurobiology of Aging, 2017, 53, 195.e7-195.e10.	3.1	46
45	Clinical and genetic analyses of familial and sporadic frontotemporal dementia patients in Southern Italy. Alzheimer's and Dementia, 2017, 13, 858-869.	0.8	24
46	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
47	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. JAMA Neurology, 2017, 74, 780.	9.0	245
48	Exome sequencing establishes a gelsolin mutation as the cause of inherited bulbarâ€onset neuropathy. Muscle and Nerve, 2017, 56, 1001-1005.	2.2	7
49	ADORA1 mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. Movement Disorders, 2017, 32, 298-299.	3.9	11
50	SLC25A46 Mutations Associated with Autosomal Recessive Cerebellar Ataxia in North African Families. Neurodegenerative Diseases, 2017, 17, 208-212.	1.4	22
51	Establishing the role of rare coding variants in known Parkinson's disease risk loci. Neurobiology of Aging, 2017, 59, 220.e11-220.e18.	3.1	15
52	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
53	Transcriptomic profiling of the human brain reveals that altered synaptic gene expression is associated with chronological aging. Scientific Reports, 2017, 7, 16890.	3.3	47
54	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. Genome Medicine, 2016, 8, 65.	8.2	20

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55	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
56	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. Neurobiology of Aging, 2016, 43, 180.e1-180.e5.	3.1	40
57	Mutation analysis of the MS4A and TREM gene clusters inÂaÂcase-control Alzheimer's disease data set. Neurobiology of Aging, 2016, 42, 217.e7-217.e13.	3.1	28
58	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
59	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
60	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. Neurobiology of Disease, 2016, 94, 55-62.	4.4	55
61	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. Neurobiology of Aging, 2016, 46, 235.e1-235.e9.	3.1	37
62	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.	6.2	333
63	A 7.5â€Mb duplication at chromosome 11q21â€11q22.3 is associated with a novel spastic ataxia syndrome. Movement Disorders, 2015, 30, 262-266.	3.9	9
64	CHCHD2 and Parkinson's disease. Lancet Neurology, The, 2015, 14, 678-679.	10.2	50
65	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
66	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	3.1	96
67	A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396.	9.0	139
68	Association of a NovelACTA1Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. JAMA Neurology, 2015, 72, 689.	9.0	35
69	Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. Human Molecular Genetics, 2015, 24, 1504-1512.	2.9	8
70	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. Lancet Neurology, The, 2015, 14, 1002-1009.	10.2	179
71	Genome-wide meta-analysis identifies six novel loci associated with habitual coffee consumption. Molecular Psychiatry, 2015, 20, 647-656.	7.9	235
72	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666.	14.8	398

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73	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
74	Genetic comorbidities in Parkinson's disease. Human Molecular Genetics, 2014, 23, 831-841.	2.9	57
75	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e311-e311.	7.6	112
76	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
77	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
78	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
79	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.	9.0	51
80	Genome-wide association study of Tourette's syndrome. Molecular Psychiatry, 2013, 18, 721-728.	7.9	161
81	Exome sequencing: an efficient diagnostic tool for complex neurodegenerative disorders. European Journal of Neurology, 2013, 20, 486-492.	3.3	25
82	mRNA expression, splicing and editing in the embryonic and adult mouse cerebral cortex. Nature Neuroscience, 2013, 16, 499-506.	14.8	130
83	Age-associated changes in gene expression in human brain and isolated neurons. Neurobiology of Aging, 2013, 34, 1199-1209.	3.1	65
84	Genome-wide association study of obsessive-compulsive disorder. Molecular Psychiatry, 2013, 18, 788-798.	7.9	312
85	Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. American Journal of Human Genetics, 2013, 92, 245-251.	6.2	120
86	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	21.4	338
87	Using Exome Sequencing to Reveal Mutations in TREM2 Presenting as a Frontotemporal Dementiaâ€“like Syndrome Without Bone Involvement. JAMA Neurology, 2013, 70, 78.	9.0	311
88	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	2.9	122
89	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. Nucleic Acids Research, 2013, 41, e88-e88.	14.5	39
90	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105.	0.8	41

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91	Age-modulated association between prefrontal NAA and the BDNF gene. International Journal of Neuropsychopharmacology, 2013, 16, 1185-1193.	2.1	5
92	Imputation of Variants from the 1000 Genomes Project Modestly Improves Known Associations and Can Identify Low-frequency Variant - Phenotype Associations Undetected by HapMap Based Imputation. PLoS ONE, 2013, 8, e64343.	2.5	61
93	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	2.9	176
94	Exome sequencing identifies a novel TRPV4 mutation in a CMT2C family. Neurology, 2012, 79, 192-194.	1.1	34
95	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882.	7.6	114
96	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Neurology, 2012, 79, 127-131.	1.1	35
97	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. Human Mutation, 2012, 33, 1708-1718.	2.5	42
98	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. Human Molecular Genetics, 2012, 21, 4094-4103.	2.9	191
99	Exome sequencing reveals an unexpected genetic cause of disease: NOTCH3 mutation in a Turkish family with Alzheimer's disease. Neurobiology of Aging, 2012, 33, 1008.e17-1008.e23.	3.1	86
100	Integration of GWAS SNPs and tissue specific expression profiling reveal discrete eQTLs for human traits in blood and brain. Neurobiology of Disease, 2012, 47, 20-28.	4.4	121
101	Allelic heterogeneity and more detailed analyses of known loci explain additional phenotypic variation and reveal complex patterns of association. Human Molecular Genetics, 2011, 20, 4082-4092.	2.9	61
102	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705.	21.4	502
103	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2011, 69, 397.	8.1	7
104	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
105	Distinct DNA methylation changes highly correlated with chronological age in the human brain. Human Molecular Genetics, 2011, 20, 1164-1172.	2.9	360
106	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569.	6.2	54
107	Genome-wide Screen Identifies rs646776 near Sortilin as a Regulator of Progranulin Levels in Human Plasma. American Journal of Human Genetics, 2010, 87, 890-897.	6.2	130
108	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. Lancet Neurology, The, 2010, 9, 978-985.	10.2	236

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109	Abundant Quantitative Trait Loci Exist for DNA Methylation and Gene Expression in Human Brain. PLoS Genetics, 2010, 6, e1000952.	3.5	722
110	Another locus, a new method. Brain, 2010, 133, 3492-3493.	7.6	2
111	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864.	8.1	1,100
112	Genetic Variability in CLU and Its Association with Alzheimer's Disease. PLoS ONE, 2010, 5, e9510.	2.5	52
113	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106
114	A simple and efficient algorithm for genomeâ€wide homozygosity analysis in disease. Molecular Systems Biology, 2009, 5, 304.	7.2	1
115	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. PLoS Genetics, 2009, 5, e1000415.	3.5	76
116	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	5.3	257
117	Extended tracts of homozygosity identify novel candidate genes associated with late-onset Alzheimerâ€™s disease. Neurogenetics, 2009, 10, 183-190.	1.4	104
118	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	21.4	1,745
119	Genetic Control of Human Brain Transcript Expression in Alzheimer Disease. American Journal of Human Genetics, 2009, 84, 445-458.	6.2	290
120	Structural genomic variation in ischemic stroke. Neurogenetics, 2008, 9, 101-108.	1.4	32
121	Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322.	2.5	46
122	Comprehensive analysis ofLRRK2in publicly available Parkinson's disease cases and neurologically normal controls. Human Mutation, 2008, 29, 485-490.	2.5	96
123	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). PLoS Genetics, 2008, 4, e1000072.	3.5	415
124	Genotype, haplotype and copy-number variation in worldwide human populations. Nature, 2008, 451, 998-1003.	27.8	780
125	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
126	RNA binding activity of the recessive parkinsonism protein DJ-1 supports involvement in multiple cellular pathways. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 10244-10249.	7.1	196

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127	Genome-wide SNP assay reveals structural genomic variation, extended homozygosity and cell-line induced alterations in normal individuals. Human Molecular Genetics, 2007, 16, 1-14.	2.9	211
128	A survey of genetic human cortical gene expression. Nature Genetics, 2007, 39, 1494-1499.	21.4	488
129	Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. Lancet Neurology, The, 2007, 6, 322-328.	10.2	206
130	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. Lancet Neurology, The, 2007, 6, 414-420.	10.2	175
131	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. Lancet Neurology, The, 2006, 5, 911-916.	10.2	360
132	Application of Genome-Wide Single Nucleotide Polymorphism Typing: Simple Association and Beyond. PLoS Genetics, 2006, 2, e150.	3.5	85
133	Association of Tau Haplotype-Tagging Polymorphisms with Parkinsonâ€™s Disease in Diverse Ethnic Parkinsonâ€™s Disease Cohorts. Neurodegenerative Diseases, 2006, 3, 327-333.	1.4	38
134	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
135	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	1
136	Mutations in the Sphingolipid Pathway Gene <i>SPTLC1</i> are a Cause of Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	0