Nabec North American Brain Expression Consortium

List of Publications by Year in Descending Order

140 papers	19,479 citations	59 h-index	139 g-index
157 ext. papers	23,569 ext. citations	11.9 avg, IF	5.21 L-index

#	Paper	IF	Citations
140	RNA sequencing of whole blood reveals early alterations in immune cells and gene expression in Parkinson's disease. *Nature Aging*, **2021**, 1, 734-747		2
139	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	4.9	2
138	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. *JAMA Neurology*, **2021**, 78, 464-472	17.2	17
137	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. *Annals of Neurology*, **2021**, 90, 35-42	9.4	6
136	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. *Movement Disorders*, **2021**, 36, 1795-1804	7	5
135	Identification of sixteen novel candidate genes for late onset Parkinson's disease. *Molecular Neurodegeneration*, **2021**, 16, 35	19	3
134	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Neuron*, **2021**, 109, 448-460.e4	13.9	20
133	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. *Lancet Neurology, The*, **2021**, 20, 107-116	24.1	23
132	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. *Neurology*, **2021**, 96, e600-e609	6.5	6
131	Replication assessment of NUS1 variants in Parkinson's disease. *Neurobiology of Aging*, **2021**, 101, 300.e1-300.e3	5.6	3
130	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. *Movement Disorders*, **2021**, 36, 449-459	7	2
129	The Parkinson's Disease DNA Variant Browser. *Movement Disorders*, **2021**, 36, 1250-1258	7	3
128	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. *Nature Genetics*, **2021**, 53, 294-303	36.3	31
127	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2021**, 78, 1236-1248	17.2	5
126	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. *Nature Communications*, **2021**, 12, 7342	17.4	2
125	Large-scale pathway specific polygenic risk and transcriptomic community network analysis identifies novel functional pathways in Parkinson disease. *Acta Neuropathologica*, **2020**, 140, 341-358	14.3	19
124	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. *Nature Communications*, **2020**, 11, 1041	17.4	6

123	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. *Movement Disorders*, **2020**, 35, 774-780	7	27
122	MIDN locus structural variants and Parkinson's Disease risk. *Annals of Clinical and Translational Neurology*, **2020**, 7, 602-603	5.3	1
121	Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. *Brain*, **2020**, 143, 234-248	11.2	69
120	A SINE-VNTR- in the LRIG2 Promoter Is Associated with Gene Expression at the Locus. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	3
119	The Parkinson's Disease Genome-Wide Association Study Locus Browser. *Movement Disorders*, **2020**, 35, 2056-2067	7	26
118	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. *Movement Disorders*, **2019**, 34, 1851-1863	7	18
117	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. *Movement Disorders*, **2019**, 34, 460-468	7	40
116	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. *Movement Disorders*, **2019**, 34, 1333-1344	7	14
115	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. *Npj Parkinsons Disease*, **2019**, 5, 8	9.7	47
114	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinson's disease heritability. *Npj Parkinsons Disease*, **2019**, 5, 6	9.7	51
113	Parkinson's disease age at onset genome-wide association study: Defining heritability, genetic loci, and □synuclein mechanisms. *Movement Disorders*, **2019**, 34, 866-875	7	136
112	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. *Annals of Neurology*, **2019**, 85, 470-481	9.4	72
111	Genomewide association study of Parkinson's disease clinical biomarkers in 12 longitudinal patients' cohorts. *Movement Disorders*, **2019**, 34, 1839-1850	7	69
110	The Parkinson's Disease Mendelian Randomization Research Portal. *Movement Disorders*, **2019**, 34, 1864-1872	7	29
109	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	24.1	562
108	Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. *JAMA Neurology*, **2018**, 75, 591-599	17.2	58
107	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. *Neurobiology of Aging*, **2018**, 64, 159.e5-159.e8	5.6	23
106	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. *Neurobiology of Aging*, **2018**, 66, 179.e17-179.e29	5.6	23

105	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
104	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. *JAMA Neurology*, **2018**, 75, 1416-1422	17.2	50
103	A comprehensive analysis of SNCA-related genetic risk in sporadic parkinson disease. *Annals of Neurology*, **2018**, 84, 117-129	9.4	33
102	Novel genetic loci associated with hippocampal volume. *Nature Communications*, **2017**, 8, 13624	17.4	173
101	Discovery and functional prioritization of Parkinson's disease candidate genes from large-scale whole exome sequencing. *Genome Biology*, **2017**, 18, 22	18.3	62
100	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	5.6	32
99	Clinical and genetic analyses of familial and sporadic frontotemporal dementia patients in Southern Italy. *Alzheimer's and Dementia*, **2017**, 13, 858-869	1.2	18
98	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	5.6	54
97	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. *JAMA Neurology*, **2017**, 74, 780-792	17.2	150
96	Exome sequencing establishes a gelsolin mutation as the cause of inherited bulbar-onset neuropathy. *Muscle and Nerve*, **2017**, 56, 1001-1005	3.4	5
95	ADORA1 mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. *Movement Disorders*, **2017**, 32, 298-299	7	10
94	Transcriptomic profiling of the human brain reveals that altered synaptic gene expression is associated with chronological aging. *Scientific Reports*, **2017**, 7, 16890	4.9	22
93	SLC25A46 Mutations Associated with Autosomal Recessive Cerebellar Ataxia in North African Families. *Neurodegenerative Diseases*, **2017**, 17, 208-212	2.3	16
92	Establishing the role of rare coding variants in known Parkinson's disease risk loci. *Neurobiology of Aging*, **2017**, 59, 220.e11-220.e18	5.6	11
91	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. *Brain*, **2017**, 140, 3191-3203	11.2	209
90	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. *Human Molecular Genetics*, **2016**, 25, 5483-5489	5.6	40
89	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. *Neurobiology of Disease*, **2016**, 94, 55-62	7.5	41
88	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. *Neurobiology of Aging*, **2016**, 46, 235.e1-9	5.6	33

87	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	11	225
86	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. *Genome Medicine*, **2016**, 8, 65	14.4	14
85	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. *PLoS ONE*, **2016**, 11, e0162592	3.7	16
84	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. *Neurobiology of Aging*, **2016**, 43, 180.e1-5	5.6	32
83	Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. *Neurobiology of Aging*, **2016**, 42, 217.e7-217.e13	5.6	24
82	Novel genetic loci underlying human intracranial volume identified through genome-wide association. *Nature Neuroscience*, **2016**, 19, 1569-1582	25.5	147
81	Common genetic variants influence human subcortical brain structures. *Nature*, **2015**, 520, 224-9	50.4	601
80	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. *Neurobiology of Aging*, **2015**, 36, 1605.e7-12	5.6	70
79	A genome-wide association study of myasthenia gravis. *JAMA Neurology*, **2015**, 72, 396-404	17.2	99
78	Association of a Novel ACTA1 Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. *JAMA Neurology*, **2015**, 72, 689-98	17.2	27
77	Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. *Human Molecular Genetics*, **2015**, 24, 1504-12	5.6	7
76	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. *Lancet Neurology, The*, **2015**, 14, 1002-9	24.1	141
75	Genome-wide meta-analysis identifies six novel loci associated with habitual coffee consumption. *Molecular Psychiatry*, **2015**, 20, 647-656	15.1	167
74	A 7.5-Mb duplication at chromosome 11q21-11q22.3 is associated with a novel spastic ataxia syndrome. *Movement Disorders*, **2015**, 30, 262-6	7	6
73	CHCHD2 and Parkinson's disease. *Lancet Neurology, The*, **2015**, 14, 678-9	24.1	43
72	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. *Nature Neuroscience*, **2014**, 17, 664-666	25.5	319
71	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. *Nature*, **2014** , 505, 550-554	50.4	345
70	Genetic comorbidities in Parkinson's disease. *Human Molecular Genetics*, **2014**, 23, 831-41	5.6	49

69	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. *Brain* , **2014**, 137, e311	11.2	89
68	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. *Nature Genetics*, **2014**, 46, 989-93	36.3	1261
67	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. *Brain*, **2014**, 137, 2480-92	11.2	127
66	Missense variant in TREML2 protects against Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 1510.e19-26	5.6	84
65	Analysis of genome-wide association studies of Alzheimer disease and of Parkinson disease to determine if these 2 diseases share a common genetic risk. *JAMA Neurology*, **2013**, 70, 1268-76	17.2	46
64	Genome-wide association study of Tourette's syndrome. *Molecular Psychiatry*, **2013**, 18, 721-8	15.1	138
63	Exome sequencing: an efficient diagnostic tool for complex neurodegenerative disorders. *European Journal of Neurology*, **2013**, 20, 486-492	6	20
62	mRNA expression, splicing and editing in the embryonic and adult mouse cerebral cortex. *Nature Neuroscience*, **2013**, 16, 499-506	25.5	107
61	Age-associated changes in gene expression in human brain and isolated neurons. *Neurobiology of Aging*, **2013**, 34, 1199-209	5.6	44
60	Genome-wide association study of obsessive-compulsive disorder. *Molecular Psychiatry*, **2013**, 18, 788-98	15.1	244
59	Mutations in GBA2 cause autosomal-recessive cerebellar ataxia with spasticity. *American Journal of Human Genetics*, **2013**, 92, 245-51	11	101
58	Genome-wide meta-analysis identifies new susceptibility loci for migraine. *Nature Genetics*, **2013**, 45, 912-917	36.3	276
57	Using exome sequencing to reveal mutations in TREM2 presenting as a frontotemporal dementia-like syndrome without bone involvement. *JAMA Neurology*, **2013**, 70, 78-84	17.2	257
56	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. *Human Molecular Genetics*, **2013**, 22, 1039-49	5.6	96
55	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. *Nucleic Acids Research*, **2013**, 41, e88	20.1	38
54	Initial assessment of the pathogenic mechanisms of the recently identified Alzheimer risk Loci. *Annals of Human Genetics*, **2013**, 77, 85-105	2.2	40
53	Age-modulated association between prefrontal NAA and the BDNF gene. *International Journal of Neuropsychopharmacology*, **2013**, 16, 1185-93	5.8	4
52	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	3.7	42

51	Integration of GWAS SNPs and tissue specific expression profiling reveal discrete eQTLs for human traits in blood and brain. *Neurobiology of Disease*, **2012**, 47, 20-8	7.5	100
50	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. *Neurology*, **2012**, 79, 127-31	6.5	29
49	Use of support vector machines for disease risk prediction in genome-wide association studies: concerns and opportunities. *Human Mutation*, **2012**, 33, 1708-18	4.7	28
48	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. *Human Molecular Genetics*, **2012**, 21, 4094-103	5.6	134
47	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.e17-23	5.6	72
46	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. *Human Molecular Genetics*, **2012**, 21, 4996-5009	5.6	145
45	Exome sequencing identifies a novel TRPV4 mutation in a CMT2C family. *Neurology*, **2012**, 79, 192-4	6.5	29
44	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. *Brain*, **2012**, 135, 2875-82	11.2	90
43	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. *Nature Genetics*, **2011**, 43, 699-705	36.3	386
42	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. *Neuron*, **2011**, 69, 397	13.9	4
41	A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9p21-linked ALS-FTD. *Neuron*, **2011**, 72, 257-68	13.9	3018
40	Allelic heterogeneity and more detailed analyses of known loci explain additional phenotypic variation and reveal complex patterns of association. *Human Molecular Genetics*, **2011**, 20, 4082-92	5.6	51
39	Distinct DNA methylation changes highly correlated with chronological age in the human brain. *Human Molecular Genetics*, **2011**, 20, 1164-72	5.6	312
38	Abundant quantitative trait loci exist for DNA methylation and gene expression in human brain. *PLoS Genetics*, **2010**, 6, e1000952	6	612
37	Another locus, a new method. *Brain*, **2010**, 133, 3492-3	11.2	2
36	Exome sequencing reveals VCP mutations as a cause of familial ALS. *Neuron*, **2010**, 68, 857-64	13.9	939
35	Exome sequencing in Brown-Vialetto-van Laere syndrome. *American Journal of Human Genetics*, **2010**, 87, 567-9; author reply 569-70	11	48
34	Genome-wide screen identifies rs646776 near sortilin as a regulator of progranulin levels in human plasma. *American Journal of Human Genetics*, **2010**, 87, 890-7	11	110

33	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. *Lancet Neurology, The*, **2010**, 9, 978-85	24.1	206
32	Genetic variability in CLU and its association with Alzheimer's disease. *PLoS ONE*, **2010**, 5, e9510	3.7	46
31	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2009**, 18, 1524-32	5.6	91
30	A simple and efficient algorithm for genome-wide homozygosity analysis in disease. *Molecular Systems Biology*, **2009**, 5, 304	12.2	1
29	Measures of autozygosity in decline: globalization, urbanization, and its implications for medical genetics. *PLoS Genetics*, **2009**, 5, e1000415	6	63
28	SNCA variants are associated with increased risk for multiple system atrophy. *Annals of Neurology*, **2009**, 65, 610-4	9.4	232
27	Extended tracts of homozygosity identify novel candidate genes associated with late-onset Alzheimer's disease. *Neurogenetics*, **2009**, 10, 183-90	3	82
26	Genome-wide association study reveals genetic risk underlying Parkinson's disease. *Nature Genetics* , **2009**, 41, 1308-12	36.3	1469
25	Genetic control of human brain transcript expression in Alzheimer disease. *American Journal of Human Genetics*, **2009**, 84, 445-58	11	229
24	Genotype, haplotype and copy-number variation in worldwide human populations. *Nature*, **2008**, 451, 998-1003	50.4	662
23	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. *Lancet Neurology, The*, **2008**, 7, 207-15	24.1	159
22	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, 10244-9	11.5	171
21	Structural genomic variation in ischemic stroke. *Neurogenetics*, **2008**, 9, 101-8	3	26
20	Genomewide SNP assay reveals mutations underlying Parkinson disease. *Human Mutation*, **2008**, 29, 315-22	4.7	44
19	Comprehensive analysis of LRRK2 in publicly available Parkinson's disease cases and neurologically normal controls. *Human Mutation*, **2008**, 29, 485-90	4.7	82
18	A genome-wide association study identifies protein quantitative trait loci (pQTLs). *PLoS Genetics*, **2008**, 4, e1000072	6	331
17	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	5.6	199
16	A survey of genetic human cortical gene expression. *Nature Genetics*, **2007**, 39, 1494-9	36.3	413

15	Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. *Lancet Neurology, The*, **2007**, 6, 322-8	24.1	172
14	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. *Lancet Neurology, The*, **2007**, 6, 414-20	24.1	154
13	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. *Lancet Neurology, The*, **2006**, 5, 911-6	24.1	323
12	Application of genome-wide single nucleotide polymorphism typing: simple association and beyond. *PLoS Genetics*, **2006**, 2, e150	6	79
11	Association of tau haplotype-tagging polymorphisms with Parkinson's disease in diverse ethnic Parkinson's disease cohorts. *Neurodegenerative Diseases*, **2006**, 3, 327-33	2.3	32
10	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. *SSRN Electronic Journal*,	1	2
9	The genetic architecture of Parkinson disease in Spain: characterizing population-specific risk, differential haplotype structures, and providing etiologic insight		1
8	The Parkinson Disease GWAS Locus Browser		1
7	Accelerating Medicines Partnership: Parkinson Disease. Genetic Resource		2
6	Expanding Parkinson disease genetics: novel risk loci, genomic context, causal insights and heritable risk		51
5	Parkinson disease age of onset GWAS: defining heritability, genetic loci and a-synuclein mechanisms		6
4	The Parkinson Disease Mendelian Randomization Research Portal		3
3	Penetrance of Parkinson disease in LRRK2 p.G2019S carriers is modified by a polygenic risk score		3
2	Quality Control Metrics for Whole Blood Transcriptome Analysis in the Parkinson Progression Markers Initiative (PPMI)		2
1	Association of a Common Genetic Variant with Parkinson Disease is Propagated through Microglia		4