

Nabec North American Brain Expression

List of Publications by Year
in descending order

Source: <https://exaly.com/author-pdf/5738633/publications.pdf>

Version: 2024-02-01

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	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. <i>Neuron</i> , 2011, 72, 257-268.	8.1	3,833
2	Genome-wide association study reveals genetic risk underlying Parkinson's disease. <i>Nature Genetics</i> , 2009, 41, 1308-1312.	21.4	1,745
3	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.	21.4	1,685
4	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.	10.2	1,414
5	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. <i>Neuron</i> , 2010, 68, 857-864.	8.1	1,100
6	Genotype, haplotype and copy-number variation in worldwide human populations. <i>Nature</i> , 2008, 451, 998-1003.	27.8	780
7	Common genetic variants influence human subcortical brain structures. <i>Nature</i> , 2015, 520, 224-229.	27.8	772
8	Abundant Quantitative Trait Loci Exist for DNA Methylation and Gene Expression in Human Brain. <i>PLoS Genetics</i> , 2010, 6, e1000952.	3.5	722
9	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
10	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. <i>Nature Genetics</i> , 2011, 43, 699-705.	21.4	502
11	A survey of genetic human cortical gene expression. <i>Nature Genetics</i> , 2007, 39, 1494-1499.	21.4	488
12	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. <i>Nature</i> , 2014, 505, 550-554.	27.8	425
13	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). <i>PLoS Genetics</i> , 2008, 4, e1000072.	3.5	415
14	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. <i>Nature Neuroscience</i> , 2014, 17, 664-666.	14.8	398
15	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology</i> , The, 2006, 5, 911-916.	10.2	360
16	Distinct DNA methylation changes highly correlated with chronological age in the human brain. <i>Human Molecular Genetics</i> , 2011, 20, 1164-1172.	2.9	360
17	Genome-wide meta-analysis identifies new susceptibility loci for migraine. <i>Nature Genetics</i> , 2013, 45, 912-917.	21.4	338
18	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	6.2	333

#	ARTICLE	IF	CITATIONS
19	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017, 140, 3191-3203.	7.6	323
20	Genome-wide association study of obsessive-compulsive disorder. <i>Molecular Psychiatry</i> , 2013, 18, 788-798.	7.9	312
21	Using Exome Sequencing to Reveal Mutations in TREM2 Presenting as a Frontotemporal Dementia-like Syndrome Without Bone Involvement. <i>JAMA Neurology</i> , 2013, 70, 78.	9.0	311
22	Genetic Control of Human Brain Transcript Expression in Alzheimer Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 445-458.	6.2	290
23	Parkinson's disease age at onset genome-wide association study: Defining heritability, genetic loci, and L-synuclein mechanisms. <i>Movement Disorders</i> , 2019, 34, 866-875.	3.9	258
24	SNCA variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009, 65, 610-614.	5.3	257
25	Novel genetic loci associated with hippocampal volume. <i>Nature Communications</i> , 2017, 8, 13624.	12.8	250
26	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. <i>JAMA Neurology</i> , 2017, 74, 780.	9.0	245
27	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. <i>Lancet Neurology</i> , The, 2010, 9, 978-985.	10.2	236
28	Genome-wide meta-analysis identifies six novel loci associated with habitual coffee consumption. <i>Molecular Psychiatry</i> , 2015, 20, 647-656.	7.9	235
29	Novel genetic loci underlying human intracranial volume identified through genome-wide association. <i>Nature Neuroscience</i> , 2016, 19, 1569-1582.	14.8	213
30	Genome-wide SNP assay reveals structural genomic variation, extended homozygosity and cell-line induced alterations in normal individuals. <i>Human Molecular Genetics</i> , 2007, 16, 1-14.	2.9	211
31	Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology</i> , The, 2007, 6, 322-328.	10.2	206
32	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. <i>Lancet Neurology</i> , The, 2008, 7, 207-215.	10.2	202
33	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. <i>Nature Genetics</i> , 2021, 53, 294-303.	21.4	198
34	RNA binding activity of the recessive parkinsonism protein DJ-1 supports involvement in multiple cellular pathways. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 10244-10249.	7.1	196
35	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. <i>Human Molecular Genetics</i> , 2012, 21, 4094-4103.	2.9	191
36	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. <i>Lancet Neurology</i> , The, 2015, 14, 1002-1009.	10.2	179

#	ARTICLE	IF	CITATIONS
37	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	2.9	176
38	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
39	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
40	Genome-wide association study of Tourette's syndrome. Molecular Psychiatry, 2013, 18, 721-728.	7.9	161
41	Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. Brain, 2020, 143, 234-248.	7.6	149
42	A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396.	9.0	139
43	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
44	mRNA expression, splicing and editing in the embryonic and adult mouse cerebral cortex. Nature Neuroscience, 2013, 16, 499-506.	14.8	130
45	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
46	Genomewide association study of Parkinson's disease clinical biomarkers in 12 longitudinal patients' cohorts. Movement Disorders, 2019, 34, 1839-1850.	3.9	122
47	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
48	Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. American Journal of Human Genetics, 2013, 92, 245-251.	6.2	120
49	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
50	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882.	7.6	114
51	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e311-e311.	7.6	112
52	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
53	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
54	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106

#	ARTICLE	IF	CITATIONS
55	Extended tracts of homozygosity identify novel candidate genes associated with late-onset Alzheimer's disease. <i>Neurogenetics</i> , 2009, 10, 183-190.	1.4	104
56	Comprehensive analysis of <i>LRRK2</i> in publicly available Parkinson's disease cases and neurologically normal controls. <i>Human Mutation</i> , 2008, 29, 485-490.	2.5	96
57	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2015, 36, 1605.e7-1605.e12.	3.1	96
58	Discovery and functional prioritization of Parkinson's disease candidate genes from large-scale whole exome sequencing. <i>Genome Biology</i> , 2017, 18, 22.	8.8	96
59	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. <i>Npj Parkinson's Disease</i> , 2019, 5, 8.	5.3	95
60	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	9.0	95
61	Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. <i>JAMA Neurology</i> , 2018, 75, 591.	9.0	93
62	Exome sequencing reveals an unexpected genetic cause of disease: NOTCH3 mutation in a Turkish family with Alzheimer's disease. <i>Neurobiology of Aging</i> , 2012, 33, 1008.e17-1008.e23.	3.1	86
63	Application of Genome-Wide Single Nucleotide Polymorphism Typing: Simple Association and Beyond. <i>PLoS Genetics</i> , 2006, 2, e150.	3.5	85
64	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinson's disease heritability. <i>Npj Parkinson's Disease</i> , 2019, 5, 6.	5.3	83
65	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. <i>PLoS Genetics</i> , 2009, 5, e1000415.	3.5	76
66	The Parkinson's Disease <sc>Genome-Wide</sc> Association Study Locus Browser. <i>Movement Disorders</i> , 2020, 35, 2056-2067.	3.9	68
67	Large-scale pathway specific polygenic risk and transcriptomic community network analysis identifies novel functional pathways in Parkinson disease. <i>Acta Neuropathologica</i> , 2020, 140, 341-358.	7.7	68
68	Frequency of Loss of Function Variants in <i>LRRK2</i> in Parkinson Disease. <i>JAMA Neurology</i> , 2018, 75, 1416.	9.0	66
69	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 460-468.	3.9	66
70	Age-associated changes in gene expression in human brain and isolated neurons. <i>Neurobiology of Aging</i> , 2013, 34, 1199-1209.	3.1	65
71	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. <i>Lancet Neurology</i> , The, 2021, 20, 107-116.	10.2	62
72	Allelic heterogeneity and more detailed analyses of known loci explain additional phenotypic variation and reveal complex patterns of association. <i>Human Molecular Genetics</i> , 2011, 20, 4082-4092.	2.9	61

#	ARTICLE	IF	CITATIONS
73	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
74	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. Movement Disorders, 2021, 36, 1795-1804.	3.9	60
75	Genetic comorbidities in Parkinson's disease. Human Molecular Genetics, 2014, 23, 831-841.	2.9	57
76	Penetrance of Parkinson's Disease in <i>LRRK2</i> p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	3.9	57
77	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
78	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. Neurobiology of Disease, 2016, 94, 55-62.	4.4	55
79	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569.	6.2	54
80	Genetic Variability in CLU and Its Association with Alzheimer's Disease. PLoS ONE, 2010, 5, e9510.	2.5	52
81	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
82	CHCHD2 and Parkinson's disease. Lancet Neurology, The, 2015, 14, 678-679.	10.2	50
83	A comprehensive analysis of <i>SNCA</i> -related genetic risk in sporadic parkinson disease. Annals of Neurology, 2018, 84, 117-129.	5.3	50
84	The Parkinson's Disease Mendelian Randomization Research Portal. Movement Disorders, 2019, 34, 1864-1872.	3.9	50
85	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. Human Molecular Genetics, 2016, 25, ddw348.	2.9	48
86	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
87	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.	3.9	47
88	Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322.	2.5	46
89	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
90	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46

#	ARTICLE	IF	CITATIONS
91	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome. <i>Nature Communications</i> , 2021, 12, 7342.	12.8	44
92	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. <i>Human Mutation</i> , 2012, 33, 1708-1718.	2.5	42
93	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. <i>Annals of Human Genetics</i> , 2013, 77, 85-105.	0.8	41
94	Identification of sixteen novel candidate genes for late onset Parkinson's disease. <i>Molecular Neurodegeneration</i> , 2021, 16, 35.	10.8	41
95	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. <i>Neurobiology of Aging</i> , 2016, 43, 180.e1-180.e5.	3.1	40
96	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. <i>Nucleic Acids Research</i> , 2013, 41, e88-e88.	14.5	39
97	Association of Tau Haplotype-Tagging Polymorphisms with Parkinson's Disease in Diverse Ethnic Parkinson's Disease Cohorts. <i>Neurodegenerative Diseases</i> , 2006, 3, 327-333.	1.4	38
98	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016, 46, 235.e1-235.e9.	3.1	37
99	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. <i>Neurology</i> , 2012, 79, 127-131.	1.1	35
100	Association of a Novel <i>ACTA1</i> Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. <i>JAMA Neurology</i> , 2015, 72, 689.	9.0	35
101	Exome sequencing identifies a novel TRPV4 mutation in a CMT2C family. <i>Neurology</i> , 2012, 79, 192-194.	1.1	34
102	Structural genomic variation in ischemic stroke. <i>Neurogenetics</i> , 2008, 9, 101-108.	1.4	32
103	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.	3.1	32
104	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. <i>Neurobiology of Aging</i> , 2018, 64, 159.e5-159.e8.	3.1	30
105	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021, 90, 35-42.	5.3	29
106	Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. <i>Neurobiology of Aging</i> , 2016, 42, 217.e7-217.e13.	3.1	28
107	Heterozygous <i>PRKN</i> mutations are common but do not increase the risk of Parkinson's disease. <i>Brain</i> , 2022, 145, 2077-2091.	7.6	26
108	Exome sequencing: an efficient diagnostic tool for complex neurodegenerative disorders. <i>European Journal of Neurology</i> , 2013, 20, 486-492.	3.3	25

#	ARTICLE	IF	CITATIONS
109	Clinical and genetic analyses of familial and sporadic frontotemporal dementia patients in Southern Italy. <i>Alzheimer's and Dementia</i> , 2017, 13, 858-869.	0.8	24
110	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. <i>Neurology</i> , 2021, 96, e600-e609.	1.1	23
111	SLC25A46 Mutations Associated with Autosomal Recessive Cerebellar Ataxia in North African Families. <i>Neurodegenerative Diseases</i> , 2017, 17, 208-212.	1.4	22
112	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. <i>Nature Communications</i> , 2020, 11, 1041.	12.8	22
113	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. <i>Movement Disorders</i> , 2019, 34, 1333-1344.	3.9	21
114	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. <i>Genome Medicine</i> , 2016, 8, 65.	8.2	20
115	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.	2.5	19
116	RNA sequencing of whole blood reveals early alterations in immune cells and gene expression in Parkinson's disease. <i>Nature Aging</i> , 2021, 1, 734-747.	11.6	18
117	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. <i>Brain</i> , 2022, 145, 1757-1762.	7.6	17
118	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. <i>Movement Disorders</i> , 2021, 36, 449-459.	3.9	16
119	ATXN2 intermediate expansions in amyotrophic lateral sclerosis. <i>Brain</i> , 2022, 145, 2671-2676.	7.6	16
120	Establishing the role of rare coding variants in known Parkinson's disease risk loci. <i>Neurobiology of Aging</i> , 2017, 59, 220.e11-220.e18.	3.1	15
121	ADORA1 mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. <i>Movement Disorders</i> , 2017, 32, 298-299.	3.9	11
122	The Parkinson's Disease DNA Variant Browser. <i>Movement Disorders</i> , 2021, 36, 1250-1258.	3.9	11
123	A 7.5Mb duplication at chromosome 11q21-q22.3 is associated with a novel spastic ataxia syndrome. <i>Movement Disorders</i> , 2015, 30, 262-266.	3.9	9
124	Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , 2015, 24, 1504-1512.	2.9	8
125	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. <i>Neuron</i> , 2011, 69, 397.	8.1	7
126	Exome sequencing establishes a gelsolin mutation as the cause of inherited bulbar-onset neuropathy. <i>Muscle and Nerve</i> , 2017, 56, 1001-1005.	2.2	7

#	ARTICLE	IF	CITATIONS
127	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
128	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
129	Age-modulated association between prefrontal NAA and the BDNF gene. International Journal of Neuropsychopharmacology, 2013, 16, 1185-1193.	2.1	5
130	<i>MIDN</i> locus structural variants and Parkinson's Disease risk. Annals of Clinical and Translational Neurology, 2020, 7, 602-603.	3.7	5
131	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
132	Replication assessment of NUS1 variants in Parkinson's disease. Neurobiology of Aging, 2021, 101, 300.e1-300.e3.	3.1	3
133	Another locus, a new method. Brain, 2010, 133, 3492-3493.	7.6	2
134	A simple and efficient algorithm for genome-wide homozygosity analysis in disease. Molecular Systems Biology, 2009, 5, 304.	7.2	1
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