J Raphael Gibbs

List of Publications by Year
in descending order

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101 papers	22,298 citations	25034 57 h-index	32842 100 g-index
117 all docs	117 docs citations	117 times ranked	29023 citing authors

#	Article	IF	Citations
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
2	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	21.4	1,745
3	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
4	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864.	8.1	1,100
5	Genotype, haplotype and copy-number variation in worldwide human populations. Nature, 2008, 451, 998-1003.	27.8	780
6	Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229.	27.8	772
7	Abundant Quantitative Trait Loci Exist for DNA Methylation and Gene Expression in Human Brain. PLoS Genetics, 2010, 6, e1000952.	3.5	722
8	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
9	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705.	21.4	502
10	A survey of genetic human cortical gene expression. Nature Genetics, 2007, 39, 1494-1499.	21.4	488
11	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
12	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). PLoS Genetics, 2008, 4, e1000072.	3.5	415
13	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666.	14.8	398
14	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
15	Distinct DNA methylation changes highly correlated with chronological age in the human brain. Human Molecular Genetics, 2011, 20, 1164-1172.	2.9	360
16	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
17	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
18	Genetic Control of Human Brain Transcript Expression in Alzheimer Disease. American Journal of Human Genetics, 2009, 84, 445-458.	6.2	290

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19	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
20	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	5.3	257
21	Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624.	12.8	250
22	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. JAMA Neurology, 2017, 74, 780.	9.0	245
23	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. Lancet Neurology, The, 2010, 9, 978-985.	10.2	236
24	Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582.	14.8	213
25	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
26	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
27	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
28	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
29	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
30	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
31	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
32	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	2.9	176
33	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
34	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	7.6	169
35	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
36	A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396.	9.0	139

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37	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
38	mRNA expression, splicing and editing in the embryonic and adult mouse cerebral cortex. Nature Neuroscience, 2013, 16, 499-506.	14.8	130
39	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
40	Genomewide association study of Parkinson's disease clinical biomarkers in 12 longitudinal patients' cohorts. Movement Disorders, 2019, 34, 1839-1850.	3.9	122
41	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
42	Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. American Journal of Human Genetics, 2013, 92, 245-251.	6.2	120
43	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
44	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882.	7.6	114
45	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e311-e311.	7.6	112
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	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	2.9	106
48	Comprehensive analysis ofLRRK2in publicly available Parkinson's disease cases and neurologically normal controls. Human Mutation, 2008, 29, 485-490.	2.5	96
49	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	3.1	96
50	Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22.	8.8	96
51	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
52	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
53	Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. JAMA Neurology, 2018, 75, 591.	9.0	93
54	Application of Genome-Wide Single Nucleotide Polymorphism Typing: Simple Association and Beyond. PLoS Genetics, 2006, 2, e150.	3.5	85

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55	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. PLoS Genetics, 2009, 5, e1000415.	3.5	76
56	The Parkinson's Disease <scp>Genomeâ€Wide</scp> Association Study Locus Browser. Movement Disorders, 2020, 35, 2056-2067.	3.9	68
57	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	9.0	66
58	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
59	Age-associated changes in gene expression in human brain and isolated neurons. Neurobiology of Aging, 2013, 34, 1199-1209.	3.1	65
60	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. Lancet Neurology, The, 2021, 20, 107-116.	10.2	62
61	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
62	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
63	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. Movement Disorders, 2021, 36, 1795-1804.	3.9	60
64	Genetic comorbidities in Parkinson's disease. Human Molecular Genetics, 2014, 23, 831-841.	2.9	57
65	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
66	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. Neurobiology of Disease, 2016, 94, 55-62.	4.4	55
67	Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569.	6.2	54
68	Genetic Variability in CLU and Its Association with Alzheimer's Disease. PLoS ONE, 2010, 5, e9510.	2.5	52
69	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
70	A comprehensive analysis of SNCAâ€related genetic risk in sporadic parkinson disease. Annals of Neurology, 2018, 84, 117-129.	5.3	50
71	The Parkinson's Disease Mendelian Randomization Research Portal. Movement Disorders, 2019, 34, 1864-1872.	3.9	50
72	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

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73	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
74	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
75	Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322.	2.5	46
76	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
77	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	12.8	44
78	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
79	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105.	0.8	41
80	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	10.8	41
81	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
82	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Neurology, 2012, 79, 127-131.	1.1	35
83	Structural genomic variation in ischemic stroke. Neurogenetics, 2008, 9, 101-108.	1.4	32
84	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	3.1	30
85	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	5.3	29
86	Heterozygous PRKN mutations are common but do not increase the risk of Parkinsonâ€™s disease. Brain, 2022, 145, 2077-2091.	7.6	26
87	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
88	SLC25A46 Mutations Associated with Autosomal Recessive Cerebellar Ataxia in North African Families. Neurodegenerative Diseases, 2017, 17, 208-212.	1.4	22
89	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. Genome Medicine, 2016, 8, 65.	8.2	20
90	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

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91	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
92	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. Brain, 2022, 145, 1757-1762.	7.6	17
93	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459.	3.9	16
94	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
95	ADORA1mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. Movement Disorders, 2017, 32, 298-299.	3.9	11
96	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
97	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
98	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
99	MIDN locus structural variants and Parkinson's Disease risk. Annals of Clinical and Translational Neurology, 2020, 7, 602-603.	3.7	5
100	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
101	Replication assessment of NUS1 variants in Parkinson's disease. Neurobiology of Aging, 2021, 101, 300.e1-300.e3.	3.1	3