# J Raphael Gibbs

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/10708084/publications.pdf

Version: 2024-02-01

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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 | 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 | Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. Lancet Neurology, The, 2021, 20, 107-116. | 10.2 | 62 |
| 4 | Replication assessment of NUS1 variants in Parkinson's disease. Neurobiology of Aging, 2021, 101, 300.e1-300.e3. | 3.1 | 3 |
| 5 | Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459. | 3.9 | 16 |
| 6 | 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 |
| 7 | 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 |
| 8 | 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 |
| 9 | Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42. | 5.3 | 29 |
| 10 | Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. Movement Disorders, 2021, 36, 1795-1804. | 3.9 | 60 |
| 11 | Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35. | 10.8 | 41 |
| 12 | 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 |
| 13 | Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342. | 12.8 | 44 |
| 14 | 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 |
| 15 | The Parkinson's Disease <scp>Genomeâ€Wide</scp> Association Study Locus Browser. Movement Disorders, 2020, 35, 2056-2067. | 3.9 | 68 |
| 16 | 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 |
| 17 | MIDN locus structural variants and Parkinson's Disease risk. Annals of Clinical and Translational Neurology, 2020, 7, 602-603. | 3.7 | 5 |
| 18 | Genomewide association study of Parkinson's disease clinical biomarkers in 12 longitudinal patients' cohorts. Movement Disorders, 2019, 34, 1839-1850. | 3.9 | 122 |

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|---|---|---|---|
| 19 | The Parkinson's Disease Mendelian Randomization Research Portal. Movement Disorders, 2019, 34, 1864-1872. | 3.9 | 50 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | 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 |
| 25 | Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481. | 5.3 | 118 |
| 26 | Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. JAMA Neurology, 2018, 75, 591. | 9.0 | 93 |
| 27 | Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8. | 3.1 | 30 |
| 28 | Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6. | 8.1 | 517 |
| 29 | Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416. | 9.0 | 66 |
| 30 | A comprehensive analysis of SNCAâ€related genetic risk in sporadic parkinson disease. Annals of Neurology, 2018, 84, 117-129. | 5.3 | 50 |
| 31 | Novel genetic loci associated with hippocampal volume. Nature Communications, 2017, 8, 13624. | 12.8 | 250 |
| 32 | Discovery and functional prioritization of Parkinsonâ€™s disease candidate genes from large-scale whole exome sequencing. Genome Biology, 2017, 18, 22. | 8.8 | 96 |
| 33 | 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 |
| 34 | 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 |
| 35 | 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 |
| 36 | Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. JAMA Neurology, 2017, 74, 780. | 9.0 | 245 |

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|---|---|---|---|
| 37 | ADORA1mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. Movement Disorders, 2017, 32, 298-299. | 3.9 | 11 |
| 38 | SLC25A46 Mutations Associated with Autosomal Recessive Cerebellar Ataxia in North African Families. Neurodegenerative Diseases, 2017, 17, 208-212. | 1.4 | 22 |
| 39 | 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 |
| 40 | Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203. | 7.6 | 323 |
| 41 | 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 |
| 42 | Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. Genome Medicine, 2016, 8, 65. | 8.2 | 20 |
| 43 | 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 |
| 44 | Novel genetic loci underlying human intracranial volume identified through genome-wide association. Nature Neuroscience, 2016, 19, 1569-1582. | 14.8 | 213 |
| 45 | 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 |
| 46 | Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. Neurobiology of Disease, 2016, 94, 55-62. | 4.4 | 55 |
| 47 | 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 |
| 48 | 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 |
| 49 | Common genetic variants influence human subcortical brain structures. Nature, 2015, 520, 224-229. | 27.8 | 772 |
| 50 | NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12. | 3.1 | 96 |
| 51 | A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396. | 9.0 | 139 |
| 52 | 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 |
| 53 | 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 |
| 54 | Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666. | 14.8 | 398 |

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|---|---|---|---|
| 55 | Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554. | 27.8 | 425 |
| 56 | Genetic comorbidities in Parkinson's disease. Human Molecular Genetics, 2014, 23, 831-841. | 2.9 | 57 |
| 57 | Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. Brain, 2014, 137, e311-e311. | 7.6 | 112 |
| 58 | Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492. | 7.6 | 169 |
| 59 | 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 |
| 60 | mRNA expression, splicing and editing in the embryonic and adult mouse cerebral cortex. Nature Neuroscience, 2013, 16, 499-506. | 14.8 | 130 |
| 61 | Age-associated changes in gene expression in human brain and isolated neurons. Neurobiology of Aging, 2013, 34, 1199-1209. | 3.1 | 65 |
| 62 | Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. American Journal of Human Genetics, 2013, 92, 245-251. | 6.2 | 120 |
| 63 | 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 |
| 64 | 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 |
| 65 | Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105. | 0.8 | 41 |
| 66 | 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 |
| 67 | Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009. | 2.9 | 176 |
| 68 | Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. Brain, 2012, 135, 2875-2882. | 7.6 | 114 |
| 69 | Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Neurology, 2012, 79, 127-131. | 1.1 | 35 |
| 70 | 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 |
| 71 | 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 |
| 72 | 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 |

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|---|---|---|---|
| 73 | 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 |
| 74 | Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. Nature Genetics, 2011, 43, 699-705. | 21.4 | 502 |
| 75 | A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268. | 8.1 | 3,833 |
| 76 | Distinct DNA methylation changes highly correlated with chronological age in the human brain. Human Molecular Genetics, 2011, 20, 1164-1172. | 2.9 | 360 |
| 77 | Exome Sequencing in Brown-Vialetto-Van Laere Syndrome. American Journal of Human Genetics, 2010, 87, 567-569. | 6.2 | 54 |
| 78 | 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 |
| 79 | Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. Lancet Neurology, The, 2010, 9, 978-985. | 10.2 | 236 |
| 80 | Abundant Quantitative Trait Loci Exist for DNA Methylation and Gene Expression in Human Brain. PLoS Genetics, 2010, 6, e1000952. | 3.5 | 722 |
| 81 | Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864. | 8.1 | 1,100 |
| 82 | Genetic Variability in CLU and Its Association with Alzheimer's Disease. PLoS ONE, 2010, 5, e9510. | 2.5 | 52 |
| 83 | A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532. | 2.9 | 106 |
| 84 | Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. PLoS Genetics, 2009, 5, e1000415. | 3.5 | 76 |
| 85 | SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614. | 5.3 | 257 |
| 86 | Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312. | 21.4 | 1,745 |
| 87 | Genetic Control of Human Brain Transcript Expression in Alzheimer Disease. American Journal of Human Genetics, 2009, 84, 445-458. | 6.2 | 290 |
| 88 | Structural genomic variation in ischemic stroke. Neurogenetics, 2008, 9, 101-108. | 1.4 | 32 |
| 89 | Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322. | 2.5 | 46 |
| 90 | Comprehensive analysis ofLRRK2in publicly available Parkinson's disease cases and neurologically normal controls. Human Mutation, 2008, 29, 485-490. | 2.5 | 96 |

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|---|---|---|---|
| 91 | A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). PLoS Genetics, 2008, 4, e1000072. | 3.5 | 415 |
| 92 | Genotype, haplotype and copy-number variation in worldwide human populations. Nature, 2008, 451, 998-1003. | 27.8 | 780 |
| 93 | 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 |
| 94 | 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 |
| 95 | 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 |
| 96 | A survey of genetic human cortical gene expression. Nature Genetics, 2007, 39, 1494-1499. | 21.4 | 488 |
| 97 | 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 |
| 98 | 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 |
| 99 | 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 |
| 100 | Application of Genome-Wide Single Nucleotide Polymorphism Typing: Simple Association and Beyond. PLoS Genetics, 2006, 2, e150. | 3.5 | 85 |
| 101 | Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , . | 0.4 | 4 |