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List of Publications by Year
in descending order

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103 papers	16,939 citations	61945 43 h-index	40954 93 g-index
126 all docs	126 docs citations	126 times ranked	19396 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.	3.8	3,833
2	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	9.4	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.	4.9	1,414
4	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864.	3.8	1,100
5	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	4.9	1,039
6	Genotype, haplotype and copy-number variation in worldwide human populations. Nature, 2008, 451, 998-1003.	13.7	780
7	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
8	Challenges in the diagnosis of Parkinson's disease. Lancet Neurology, The, 2021, 20, 385-397.	4.9	468
9	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). PLoS Genetics, 2008, 4, e1000072.	1.5	415
10	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.	4.9	360
11	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	1.5	269
12	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.	2.2	258
13	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	2.8	257
14	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. Lancet Neurology, The, 2010, 9, 978-985.	4.9	236
15	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.	1.4	211
16	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.	4.9	206
17	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.	4.9	202
18	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. Nature Genetics, 2021, 53, 294-303.	9.4	198

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19	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. Lancet Neurology, The, 2018, 17, 64-74.	4.9	195
20	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. Lancet Neurology, The, 2007, 6, 414-420.	4.9	175
21	Genetic modifiers of risk and age at onset in GBA associated Parkinsonâ€™s disease and Lewy body dementia. Brain, 2020, 143, 234-248.	3.7	149
22	A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396.	4.5	139
23	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.5	139
24	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	2.8	118
25	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.	1.5	108
26	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	1.4	106
27	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. JAMA Neurology, 2021, 78, 464.	4.5	95
28	A nonsynonymous mutation in PLCG2 reduces the risk of Alzheimerâ€™s disease, dementia with Lewy bodies and frontotemporal dementia, and increases the likelihood of longevity. Acta Neuropathologica, 2019, 138, 237-250.	3.9	87
29	Human Herpesvirus 6 Detection in Alzheimerâ€™s Disease Cases and Controls across Multiple Cohorts. Neuron, 2020, 105, 1027-1035.e2.	3.8	87
30	Valosin-containing protein (VCP) mutations in sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 2231.e1-2231.e6.	1.5	86
31	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. PLoS Genetics, 2009, 5, e1000415.	1.5	76
32	A critique of the second consensus criteria for multiple system atrophy. Movement Disorders, 2019, 34, 975-984.	2.2	73
33	Parkin and PINK1 mutations in early-onset Parkinson's disease: comprehensive screening in publicly available cases and control. Journal of Medical Genetics, 2009, 46, 375-381.	1.5	72
34	Large-scale pathway specific polygenic risk and transcriptomic community network analysis identifies novel functional pathways in Parkinson disease. Acta Neuropathologica, 2020, 140, 341-358.	3.9	68
35	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	4.5	66
36	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. Movement Disorders, 2019, 34, 460-468.	2.2	66

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37	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. Lancet Neurology, The, 2021, 20, 107-116.	4.9	62
38	GLUCOCEREBROSIDASE MUTATIONS IN 108 NEUROPATHOLOGICALLY CONFIRMED CASES OF MULTIPLE SYSTEM ATROPHY. Neurology, 2009, 72, 1185-1186.	1.5	60
39	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. Movement Disorders, 2021, 36, 1795-1804.	2.2	60
40	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. Movement Disorders, 2020, 35, 774-780.	2.2	57
41	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
42	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. Neurobiology of Disease, 2016, 94, 55-62.	2.1	55
43	Conflicting Results Regarding the Semaphorin Gene (SEMA5A) and the Risk for Parkinson Disease. American Journal of Human Genetics, 2006, 78, 1082-1083.	2.6	50
44	POLG1 polyglutamine tract variants associated with Parkinson's disease. Neuroscience Letters, 2010, 477, 1-5.	1.0	47
45	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.	2.2	47
46	Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322.	1.1	46
47	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	5.8	44
48	Genome-wide estimate of the heritability of Multiple System Atrophy. Parkinsonism and Related Disorders, 2016, 22, 35-41.	1.1	42
49	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	4.4	41
50	Genetic players in multiple system atrophy: unfolding the nature of the beast. Neurobiology of Aging, 2011, 32, 1924.e5-1924.e14.	1.5	39
51	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Neurology, 2012, 79, 127-131.	1.5	35
52	Structural genomic variation in ischemic stroke. Neurogenetics, 2008, 9, 101-108.	0.7	32
53	Can Autonomic Testing and Imaging Contribute to the Early Diagnosis of Multiple System Atrophy? A Systematic Review and Recommendations by the <scp>Movement Disorder Society</scp> Multiple System Atrophy Study Group. Movement Disorders Clinical Practice, 2020, 7, 750-762.	0.8	31
54	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	1.5	30

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55	Heritability and genetic variance of dementia with Lewy bodies. Neurobiology of Disease, 2019, 127, 492-501.	2.1	29
56	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	2.8	29
57	Multiple system atrophy is not caused by C9orf72 hexanucleotide repeat expansions. Neurobiology of Aging, 2015, 36, 1223.e1-1223.e2.	1.5	25
58	Genetic analysis of neurodegenerative diseases in a pathology cohort. Neurobiology of Aging, 2019, 76, 214.e1-214.e9.	1.5	25
59	Evaluation of [123I]IBZM pinhole SPECT for the detection of striatal dopamine D2 receptor availability in rats. NeuroImage, 2005, 24, 822-831.	2.1	24
60	Genomics and Bioinformatics of Parkinson's Disease. Cold Spring Harbor Perspectives in Medicine, 2012, 2, a009449-a009449.	2.9	24
61	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. Neurology, 2021, 96, e600-e609.	1.5	23
62	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. Nature Communications, 2020, 11, 1041.	5.8	22
63	Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. International Journal of Molecular Sciences, 2015, 16, 24629-24655.	1.8	21
64	Assessment of APOE in atypical parkinsonism syndromes. Neurobiology of Disease, 2019, 127, 142-146.	2.1	21
65	Multiple system atrophy as emerging template for accelerated drug discovery in Î±-synucleinopathies. Parkinsonism and Related Disorders, 2014, 20, 793-799.	1.1	18
66	ARSA variants in Î±-synucleinopathies. Brain, 2019, 142, e70-e70.	3.7	17
67	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. Brain, 2022, 145, 1757-1762.	3.7	17
68	Mutational analysis of parkin and PINK1 in multiple system atrophy. Neurobiology of Aging, 2011, 32, 548.e5-548.e7.	1.5	16
69	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459.	2.2	16
70	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	4.9	15
71	The human prion gene M129V polymorphism is not associated with idiopathic Parkinson's disease in three distinct populations. Neuroscience Letters, 2006, 395, 227-229.	1.0	14
72	Glucocerebrosidase mutations are not found in association with LRRK2 G2019S in subjects with parkinsonism. Neuroscience Letters, 2006, 404, 163-165.	1.0	14

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73	A comprehensive screening of copy number variability in dementia with Lewy bodies. Neurobiology of Aging, 2019, 75, 223.e1-223.e10.	1.5	13
74	ADORA1mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. Movement Disorders, 2017, 32, 298-299.	2.2	11
75	<scp>Î±â€Synuclein</scp> Deposition in Sympathetic Nerve Fibers in Genetic Forms of Parkinson's Disease. Movement Disorders, 2021, 36, 2346-2357.	2.2	11
76	MAPT p.V363I mutation. Neurology: Genetics, 2019, 5, e347.	0.9	10
77	GBA mutations and Parkinson disease: When genotype meets phenotype. Neurology, 2015, 84, 866-867.	1.5	9
78	<i>C9orf72</i> Hexanucleotide Repeat Analysis in Cases with Pathologically Confirmed Dementia with Lewy Bodies. Neurodegenerative Diseases, 2016, 16, 370-372.	0.8	8
79	Classification of GBA Variants and Their Effects in Synucleinopathies. Movement Disorders, 2019, 34, 1581-1582.	2.2	8
80	LRRK2 mutations in a clinic-based cohort of Parkinson's disease. European Journal of Neurology, 2006, 13, 1298-1301.	1.7	7
81	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2011, 69, 397.	3.8	7
82	TUBB2B Mutation in an Adult Patient with Myoclonus-Dystonia. Case Reports in Neurology, 2017, 9, 216-221.	0.3	6
83	Brachial plexitis preceding encephalomyelitis in a patient with West Nile virus infection. BMJ Case Reports, 2013, 2013, bcr2013200833-bcr2013200833.	0.2	6
84	<scp>GRN</scp> Mutations Are Associated with Lewy Body Dementia. Movement Disorders, 2022, 37, 1943-1948.	2.2	5
85	Variability in clinical phenotypes of heterozygous and homozygous cases ofParkin-related Parkinson's disease. International Journal of Neuroscience, 2013, 123, 847-849.	0.8	4
86	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
87	Prion genotypes in Central America suggest selection for the V129 allele. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 33-35.	1.1	3
88	Highlighting the clinical potential of HTT repeat expansions in Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 1947-1948.	3.8	3
89	A multiplex pedigree with pathologically confirmed multiple system atrophy and Parkinsonâ€™s disease with dementia. Brain Communications, 2022, 4, .	1.5	3
90	Susceptibility genes in movement disorders. Movement Disorders, 2008, 23, 927-934.	2.2	2

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91	Restless legs syndrome: is it all in the genes?. Lancet Neurology, The, 2017, 16, 859-860.	4.9	2
92	Identification of new Î±â€synuclein regulator by nontraditional drug development pipeline. Movement Disorders, 2018, 33, 402-402.	2.2	2
93	Conjugal multiple system atrophy: Rethinking numbers of probability. Parkinsonism and Related Disorders, 2020, 77, 176-177.	1.1	1
94	Hot Topic: Epigenetics in Parkinson's Disease: A New Frontier for Diseaseâ€Modifying Therapies. Movement Disorders, 2021, 36, 862-862.	2.2	1
95	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	1
96	PINK1mutations: Does the dosage make the poison?. Human Mutation, 2009, 30, v-v.	1.1	0
97	Author response: A genome-wide association study in multiple system atrophy. Neurology, 2017, 88, 1296-1297.	1.5	0
98	O5â€04â€01: A RARE GENETIC VARIANT IN THE PLCG2 GENE IS ASSOCIATED WITH A REDUCED RISK OF ALL MAJOR TYPES OF DEMENTIA AND AN INCREASED RISK TO REACH AN EXTREMELY OLD AGE. Alzheimer's and Dementia, 2018, 14, P1648.	0.4	0
99	DNA typos spell trouble: Somatic mutations as a cause of idiopathic neurodegenerative diseases?. Movement Disorders, 2019, 34, 321-321.	2.2	0
100	Deletion at ITPR1 underlies ataxia in mice and humans (SCA15). PLoS Genetics, 2005, preprint, e108.	1.5	0
101	A Nonsynonymous Mutation in PLCG2 Reduces the Risk of Alzheimer's Disease, Dementia with Lewy-Bodies and Frontotemporal Dementia, and Increases the Likelihood of Longevity. SSRN Electronic Journal, 0, , .	0.4	0
102	Mutations in the Sphingolipid Pathway Gene <i>SPTLC1</i> are a Cause of Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	0
103	Human Herpesvirus-6 (HHV-6) Detection in Alzheimer's Disease Cases and Controls Across Multiple Cohorts. SSRN Electronic Journal, 0, , .	0.4	0