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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	Genetic evaluation of dementia with Lewy bodies implicates distinct disease subgroups. Brain, 2022, 145, 1757-1762.	3.7	17
2	<scp>GRN</scp> Mutations Are Associated with Lewy Body Dementia. Movement Disorders, 2022, 37, 1943-1948.	2.2	5
3	A multiplex pedigree with pathologically confirmed multiple system atrophy and Parkinsonâ€™s disease with dementia. Brain Communications, 2022, 4, .	1.5	3
4	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
5	Hot Topic: Epigenetics in Parkinson's Disease: A New Frontier for Diseaseâ€Modifying Therapies. Movement Disorders, 2021, 36, 862-862.	2.2	1
6	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. Lancet Neurology, The, 2021, 20, 107-116.	4.9	62
7	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. Neurology, 2021, 96, e600-e609.	1.5	23
8	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. Movement Disorders, 2021, 36, 449-459.	2.2	16
9	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
10	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
11	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	2.8	29
12	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. Movement Disorders, 2021, 36, 1795-1804.	2.2	60
13	Challenges in the diagnosis of Parkinson's disease. Lancet Neurology, The, 2021, 20, 385-397.	4.9	468
14	Highlighting the clinical potential of HTT repeat expansions in Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 1947-1948.	3.8	3
15	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	4.4	41
16	<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
17	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	5.8	44
18	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

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19	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
20	Conjugal multiple system atrophy: Rethinking numbers of probability. Parkinsonism and Related Disorders, 2020, 77, 176-177.	1.1	1
21	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
22	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. Nature Communications, 2020, 11, 1041.	5.8	22
23	Human Herpesvirus 6 Detection in Alzheimerâ€™s Disease Cases and Controls across Multiple Cohorts. Neuron, 2020, 105, 1027-1035.e2.	3.8	87
24	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
25	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
26	ARSA variants in Î±-synucleinopathies. Brain, 2019, 142, e70-e70.	3.7	17
27	Classification of GBA Variants and Their Effects in Synucleinopathies. Movement Disorders, 2019, 34, 1581-1582.	2.2	8
28	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
29	MAPT p.V363I mutation. Neurology: Genetics, 2019, 5, e347.	0.9	10
30	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
31	DNA typos spell trouble: Somatic mutations as a cause of idiopathic neurodegenerative diseases?. Movement Disorders, 2019, 34, 321-321.	2.2	0
32	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
33	A critique of the second consensus criteria for multiple system atrophy. Movement Disorders, 2019, 34, 975-984.	2.2	73
34	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
35	Heritability and genetic variance of dementia with Lewy bodies. Neurobiology of Disease, 2019, 127, 492-501.	2.1	29
36	Assessment of APOE in atypical parkinsonism syndromes. Neurobiology of Disease, 2019, 127, 142-146.	2.1	21

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37	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	2.8	118
38	Genetic analysis of neurodegenerative diseases in a pathology cohort. Neurobiology of Aging, 2019, 76, 214.e1-214.e9.	1.5	25
39	A comprehensive screening of copy number variability in dementia with Lewy bodies. Neurobiology of Aging, 2019, 75, 223.e1-223.e10.	1.5	13
40	Identification of new Î±â€synuclein regulator by nontraditional drug development pipeline. Movement Disorders, 2018, 33, 402-402.	2.2	2
41	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 2018, 64, 159.e5-159.e8.	1.5	30
42	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
43	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
44	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
45	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	4.9	15
46	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	4.5	66
47	Author response: A genome-wide association study in multiple system atrophy. Neurology, 2017, 88, 1296-1297.	1.5	0
48	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
49	ADORA1mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. Movement Disorders, 2017, 32, 298-299.	2.2	11
50	TUBB2B Mutation in an Adult Patient with Myoclonus-Dystonia. Case Reports in Neurology, 2017, 9, 216-221.	0.3	6
51	Restless legs syndrome: is it all in the genes?. Lancet Neurology, The, 2017, 16, 859-860.	4.9	2
52	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.5	139
53	<i>C9orf72</i> Hexanucleotide Repeat Analysis in Cases with Pathologically Confirmed Dementia with Lewy Bodies. Neurodegenerative Diseases, 2016, 16, 370-372.	0.8	8
54	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. Neurobiology of Disease, 2016, 94, 55-62.	2.1	55

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55	Genome-wide estimate of the heritability of Multiple System Atrophy. Parkinsonism and Related Disorders, 2016, 22, 35-41.	1.1	42
56	Genetics Underlying Atypical Parkinsonism and Related Neurodegenerative Disorders. International Journal of Molecular Sciences, 2015, 16, 24629-24655.	1.8	21
57	GBA mutations and Parkinson disease: When genotype meets phenotype. Neurology, 2015, 84, 866-867.	1.5	9
58	Multiple system atrophy is not caused by C9orf72 hexanucleotide repeat expansions. Neurobiology of Aging, 2015, 36, 1223.e1-1223.e2.	1.5	25
59	A Genome-Wide Association Study of Myasthenia Gravis. JAMA Neurology, 2015, 72, 396.	4.5	139
60	Multiple system atrophy as emerging template for accelerated drug discovery in Î±-synucleinopathies. Parkinsonism and Related Disorders, 2014, 20, 793-799.	1.1	18
61	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
62	Brachial plexitis preceding encephalomyelitis in a patient with West Nile virus infection. BMJ Case Reports, 2013, 2013, bcr2013200833-bcr2013200833.	0.2	6
63	Genomics and Bioinformatics of Parkinson's Disease. Cold Spring Harbor Perspectives in Medicine, 2012, 2, a009449-a009449.	2.9	24
64	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Neurology, 2012, 79, 127-131.	1.5	35
65	Valosin-containing protein (VCP) mutations in sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 2231.e1-2231.e6.	1.5	86
66	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
67	Mutational analysis of parkin and PINK1 in multiple system atrophy. Neurobiology of Aging, 2011, 32, 548.e5-548.e7.	1.5	16
68	Genetic players in multiple system atrophy: unfolding the nature of the beast. Neurobiology of Aging, 2011, 32, 1924.e5-1924.e14.	1.5	39
69	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2011, 69, 397.	3.8	7
70	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	3.8	3,833
71	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. Lancet Neurology, The, 2010, 9, 978-985.	4.9	236
72	POLG1 polyglutamine tract variants associated with Parkinson's disease. Neuroscience Letters, 2010, 477, 1-5.	1.0	47

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73	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. Neuron, 2010, 68, 857-864.	3.8	1,100
74	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2009, 18, 1524-1532.	1.4	106
75	Measures of Autozygosity in Decline: Globalization, Urbanization, and Its Implications for Medical Genetics. PLoS Genetics, 2009, 5, e1000415.	1.5	76
76	GLUCOCEREBROSIDASE MUTATIONS IN 108 NEUROPATHOLOGICALLY CONFIRMED CASES OF MULTIPLE SYSTEM ATROPHY. Neurology, 2009, 72, 1185-1186.	1.5	60
77	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	2.8	257
78	PINK1mutations: Does the dosage make the poison?. Human Mutation, 2009, 30, v-v.	1.1	0
79	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	9.4	1,745
80	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
81	Structural genomic variation in ischemic stroke. Neurogenetics, 2008, 9, 101-108.	0.7	32
82	Susceptibility genes in movement disorders. Movement Disorders, 2008, 23, 927-934.	2.2	2
83	Genomewide SNP assay reveals mutations underlying Parkinson disease. Human Mutation, 2008, 29, 315-322.	1.1	46
84	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). PLoS Genetics, 2008, 4, e1000072.	1.5	415
85	Genotype, haplotype and copy-number variation in worldwide human populations. Nature, 2008, 451, 998-1003.	13.7	780
86	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
87	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	1.5	269
88	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
89	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
90	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

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91	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
92	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
93	Glucocerebrosidase mutations are not found in association with LRRK2 G2019S in subjects with parkinsonism. Neuroscience Letters, 2006, 404, 163-165.	1.0	14
94	LRRK2 mutations in a clinic-based cohort of Parkinson's disease. European Journal of Neurology, 2006, 13, 1298-1301.	1.7	7
95	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
96	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
97	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
98	Deletion at ITPR1 underlies ataxia in mice and humans (SCA15). PLoS Genetics, 2005, preprint, e108.	1.5	0
99	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
100	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. SSRN Electronic Journal, 0, , .	0.4	1
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