Nicholas W. Wood

List of Publications by Year
in descending order

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472	68,214	729	906
papers	citations	120 h-index	241 g-index

499	499	499	57134
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. Science, 2004, 304, 1158-1160.	6.0	3,060
2	Second consensus statement on the diagnosis of multiple system atrophy. Neurology, 2008, 71, 670-676.	1.5	2,720
3	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	13.7	2,400
4	Cloning of the Gene Containing Mutations that Cause PARK8-Linked Parkinson's Disease. Neuron, 2004, 44, 595-600.	3.8	2,183
5	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	9.4	1,745
6	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. Nature Genetics, 2014, 46, 989-993.	9.4	1,685
7	Association between Early-Onset Parkinson's Disease and Mutations in theParkinGene. New England Journal of Medicine, 2000, 342, 1560-1567.	13.9	1,448
8	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
9	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	9.4	1,395
10	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. Lancet Neurology, The, 2008, 7, 583-590.	4.9	1,340
11	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
12	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. Nature Genetics, 2010, 42, 985-990.	9.4	918
13	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
14	Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. Lancet, The, 2011, 377, 641-649.	6.3	845
15	Interaction between ERAP1 and HLA-B27 in ankylosing spondylitis implicates peptide handling in the mechanism for HLA-B27 in disease susceptibility. Nature Genetics, 2011, 43, 761-767.	9.4	778
16	Expanding insights of mitochondrial dysfunction in Parkinson's disease. Nature Reviews Neuroscience, 2006, 7, 207-219.	4.9	773
17	Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion. Nature Genetics, 1997, 17, 65-70.	9.4	758
18	Direct Observation of the Interconversion of Normal and Toxic Forms of Î±-Synuclein. Cell, 2012, 149, 1048-1059.	13.5	755

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19	Association of Multidrug Resistance in Epilepsy with a Polymorphism in the Drug-Transporter GeneABCB1. New England Journal of Medicine, 2003, 348, 1442-1448.	13.9	690
20	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. Cell, 2018, 173, 1705-1715.e16.	13.5	623
21	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. Brain, 2009, 132, 1783-1794.	3.7	612
22	PINK1-Associated Parkinson's Disease Is Caused by Neuronal Vulnerability to Calcium-Induced Cell Death. Molecular Cell, 2009, 33, 627-638.	4.5	584
23	A robust model for read count data in exome sequencing experiments and implications for copy number variant calling. Bioinformatics, 2012, 28, 2747-2754.	1.8	534
24	Genome, transcriptome and proteome: the rise of omics data and their integration in biomedical sciences. Briefings in Bioinformatics, 2018, 19, 286-302.	3.2	498
25	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	1.5	495
26	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
27	The spectrum of pathological involvement of the striatonigral and olivopontocerebellar systems in multiple system atrophy: clinicopathological correlations. Brain, 2004, 127, 2657-2671.	3.7	493
28	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. Nature Genetics, 2009, 41, 1330-1334.	9.4	483
29	Glucocerebrosidase deficiency in substantia nigra of parkinson disease brains. Annals of Neurology, 2012, 72, 455-463.	2.8	473
30	Localization of a Novel Locus for Autosomal Recessive Early-Onset Parkinsonism, PARK6, on Human Chromosome 1p35-p36. American Journal of Human Genetics, 2001, 68, 895-900.	2.6	459
31	The mitochondrial protease HtrA2 is regulated by Parkinson's disease-associated kinase PINK1. Nature Cell Biology, 2007, 9, 1243-1252.	4.6	441
32	Parkin disease: a phenotypic study of a large case series. Brain, 2003, 126, 1279-1292.	3.7	427
33	The expression of DJ-1 (PARK7) in normal human CNS and idiopathic Parkinson's disease. Brain, 2004, 127, 420-430.	3.7	404
34	Characterization of PLA2G6 as a locus for dystoniaâ€parkinsonism. Annals of Neurology, 2009, 65, 19-23.	2.8	399
35	A common LRRK2 mutation in idiopathic Parkinson's disease. Lancet, The, 2005, 365, 415-416.	6.3	391
36	Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. Nature Genetics, 2011, 43, 117-120.	9.4	390

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37	PINK1 cleavage at position A103 by the mitochondrial protease PARL. Human Molecular Genetics, 2011, 20, 867-879.	1.4	385
38	Structural characterization of toxic oligomers that are kinetically trapped during Î±-synuclein fibril formation. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E1994-2003.	3.3	384
39	Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke. Nature Genetics, 2012, 44, 328-333.	9.4	375
40	The role of pathogenicDJ-1 mutations in Parkinson's disease. Annals of Neurology, 2003, 54, 283-286.	2.8	362
41	A conserved sorting-associated protein is mutant in chorea-acanthocytosis. Nature Genetics, 2001, 28, 119-120.	9.4	357
42	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
43	Mitochondrial function and morphology are impaired in parkinâ€mutant fibroblasts. Annals of Neurology, 2008, 64, 555-565.	2.8	339
44	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.	2.6	333
45	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
46	Genetic predictors of the maximum doses patients receive during clinical use of the anti-epileptic drugs carbamazepine and phenytoin. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 5507-5512.	3.3	321
47	Mutations in the gene LRRK2 encoding dardarin (PARK8) cause familial Parkinson's disease: clinical, pathological, olfactory and functional imaging and genetic data. Brain, 2005, 128, 2786-2796.	3.7	315
48	Targeting amyloid-Î² in glaucoma treatment. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 13444-13449.	3.3	315
49	A novel mutation in the human voltage-gated potassium channel gene (Kv1.1) associates with episodic ataxia type 1 and sometimes with partial epilepsy. Brain, 1999, 122, 817-825.	3.7	314
50	Riluzole treatment, survival and diagnostic criteria in Parkinson plus disorders: The NNIPPS Study. Brain, 2009, 132, 156-171.	3.7	298
51	The Parkinson's diseaseâ€"linked proteins Fbxo7 and Parkin interact to mediate mitophagy. Nature Neuroscience, 2013, 16, 1257-1265.	7.1	292
52	PINK1 protein in normal human brain and Parkinson's disease. Brain, 2006, 129, 1720-1731.	3.7	291
53	Parkin mutations are frequent in patients with isolated earlyâ€onset parkinsonism. Brain, 2003, 126, 1271-1278.	3.7	279
54	Understanding the molecular causes of Parkinson's disease. Trends in Molecular Medicine, 2006, 12, 521-528.	3.5	274

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55	PINK1 Is Necessary for Long Term Survival and Mitochondrial Function in Human Dopaminergic Neurons. PLoS ONE, 2008, 3, e2455.	1.1	273
56	How much phenotypic variation can be attributed toparkingenotype?. Annals of Neurology, 2003, 54, 176-185.	2.8	271
57	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	1.5	269
58	Alpha-Synuclein Oligomers Interact with Metal Ions to Induce Oxidative Stress and Neuronal Death in Parkinson's Disease. Antioxidants and Redox Signaling, 2016, 24, 376-391.	2.5	266
59	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	2.8	257
60	Mutations in the Gene PRRT2 Cause Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions. Cell Reports, 2012, 1, 2-12.	2.9	250
61	Gluten ataxia in perspective: epidemiology, genetic susceptibility and clinical characteristics. Brain, 2003, 126, 685-691.	3.7	248
62	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. PLoS Genetics, 2011, 7, e1002142.	1.5	247
63	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. JAMA Neurology, 2017, 74, 780.	4.5	245
64	Rare Deletions at 16p13.11 Predispose to a Diverse Spectrum of Sporadic Epilepsy Syndromes. American Journal of Human Genetics, 2010, 86, 707-718.	2.6	231
65	Linkage disequilibrium fine mapping and haplotype association analysis of the tau gene in progressive supranuclear palsy and corticobasal degeneration. Journal of Medical Genetics, 2005, 42, 837-846.	1.5	225
66	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 2012, 91, 1041-1050.	2.6	224
67	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	9.4	223
68	Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21. Human Molecular Genetics, 2011, 20, 345-353.	1.4	202
69	Homozygous deletions in parkin gene in European and North African families with autosomal recessive juvenile parkinsonism. Lancet, The, 1998, 352, 1355-1356.	6.3	199
70	Pathological inclusion bodies in tauopathies contain distinct complements of tau with three or four microtubule-binding repeat domains as demonstrated by new specific monoclonal antibodies. Neuropathology and Applied Neurobiology, 2003, 29, 288-302.	1.8	194
71	Huntington's disease progression. Brain, 1999, 122, 2353-2363.	3.7	193
72	Complex relationship between Parkin mutations and Parkinson disease. American Journal of Medical Genetics Part A, 2002, 114, 584-591.	2.4	193

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73	Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: An18F-dopa PET study. Annals of Neurology, 2002, 52, 849-853.	2.8	192
74	Molecular pathogenesis of Parkinson's disease. Human Molecular Genetics, 2005, 14, 2749-2755.	1.4	187
75	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	9.4	186
76	Mutations in TTBK2, encoding a kinase implicated in tau phosphorylation, segregate with spinocerebellar ataxia type 11. Nature Genetics, 2007, 39, 1434-1436.	9.4	185
77	A Genetically Mediated Bias in Decision Making Driven by Failure of Amygdala Control. Journal of Neuroscience, 2009, 29, 5985-5991.	1.7	183
78	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Annals of Neurology, 2016, 79, 983-990.	2.8	183
79	Selection and Evaluation of Tagging SNPs in the Neuronal-Sodium-Channel Gene SCN1A: Implications for Linkage-Disequilibrium Gene Mapping. American Journal of Human Genetics, 2003, 73, 551-565.	2.6	181
80	Kinetic model of the aggregation of alpha-synuclein provides insights into prion-like spreading. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E1206-15.	3.3	181
81	Mitophagy and Parkinson's disease: The PINK1â€“parkin link. Biochimica Et Biophysica Acta - Molecular Cell Research, 2011, 1813, 623-633.	1.9	176
82	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
83	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. Lancet Neurology, The, 2007, 6, 970-980.	4.9	175
84	Transcriptional repression of p53 by parkin and impairment by mutations associated with autosomal recessive juvenile Parkinson's disease. Nature Cell Biology, 2009, 11, 1370-1375.	4.6	173
85	Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. European Journal of Human Genetics, 2002, 10, 773-781.	1.4	172
86	The fragile X tremor ataxia syndrome in the differential diagnosis of multiple system atrophy: data from the EMSA Study Group. Brain, 2005, 128, 1855-1860.	3.7	172
87	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. Brain, 2016, 139, 1904-1918.	3.7	170
88	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	3.7	169
89	Mutations in the HSP27 (HSPB1) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. Neurology, 2008, 71, 1660-1668.	1.5	168
90	Common variants at the MHC locus and at chromosome 16q24.1 predispose to Barrett's esophagus. Nature Genetics, 2012, 44, 1131-1136.	9.4	162

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91	The phenotypic spectrum of DYT24 due to ANO3 mutations. Movement Disorders, 2014, 29, 928-934.	2.2	161
92	Trinucleotide repeats and neurodegenerative disease. Brain, 2004, 127, 2385-2405.	3.7	160
93	PRRT2 gene mutations. Neurology, 2012, 79, 2115-2121.	1.5	159
94	Mitochondrial dysfunction triggered by loss of HtrA2 results in the activation of a brain-specific transcriptional stress response. Cell Death and Differentiation, 2009, 16, 449-464.	5.0	156
95	Genome-Wide Association Study Implicates HLA-C*01:02 as a Risk Factor at the Major Histocompatibility Complex Locus in Schizophrenia. Biological Psychiatry, 2012, 72, 620-628.	0.7	156
96	Dopa-responsive dystonia: A clinical and molecular genetic study. Annals of Neurology, 1998, 44, 649-656.	2.8	153
97	Autosomal-dominant GTPCH1-deficient DRD: clinical characteristics and long-term outcome of 34 patients. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 839-845.	0.9	153
98	Estimating the causal influence of body mass index on risk of Parkinson disease: A Mendelian randomisation study. PLoS Medicine, 2017, 14, e1002314.	3.9	152
99	Genome scans and candidate gene approaches in the study of common diseases and variable drug responses. Trends in Genetics, 2003, 19, 615-622.	2.9	151
100	Mutations in HPCA Cause Autosomal-Recessive Primary Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 657-665.	2.6	151
101	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. PLoS Medicine, 2016, 13, e1001976.	3.9	150
102	A heterozygous effect for PINK1 mutations in Parkinson's disease?. Annals of Neurology, 2006, 60, 414-419.	2.8	149
103	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
104	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	2.8	148
105	Altered cleavage and localization of PINK1 to aggresomes in the presence of proteasomal stress. Journal of Neurochemistry, 2006, 98, 156-169.	2.1	146
106	Cortical Î±-synuclein load is associated with amyloid-Î² plaque burden in a subset of Parkinsonâ€™s disease patients. Acta Neuropathologica, 2008, 115, 417-425.	3.9	146
107	Parkinson's disease and cancer: two wars, one front. Nature Reviews Cancer, 2011, 11, 813-823.	12.8	146
108	Genetic Variants of the Î±-Synuclein Gene SNCA Are Associated with Multiple System Atrophy. PLoS ONE, 2009, 4, e7114.	1.1	144

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109	Myoclonus-dystonia syndrome: Îµ-sarcoglycan mutations and phenotype. Annals of Neurology, 2002, 52, 489-492.	2.8	143
110	A single-nucleotide polymorphism tagging set for human drug metabolism and transport. Nature Genetics, 2005, 37, 84-89.	9.4	142
111	Progression of nigrostriatal dysfunction in a parkin kindred: an [18F]dopa PET and clinical study. Brain, 2002, 125, 2248-2256.	3.7	141
112	Different patterns of electrophysiological deficits in manifesting and non-manifesting carriers of the DYT1 gene mutation. Brain, 2003, 126, 2074-2080.	3.7	141
113	Cancer and Neurodegeneration: Between the Devil and the Deep Blue Sea. PLoS Genetics, 2010, 6, e1001257.	1.5	141
114	Systematic Review and UKâ€Based Study of PARK2 (parkin), PINK1, PARK7 (DJâ€1) and LRRK2 in earlyâ€onset Parkinson's disease. Movement Disorders, 2012, 27, 1522-1529.	2.2	141
115	Autosomal Dominant Cerebellar Ataxia Type III: Linkage in a Large British Family to a 7.6-cM Region on Chromosome 15q14-21.3. American Journal of Human Genetics, 1999, 65, 420-426.	2.6	140
116	ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2015, 85, 80-88.	1.5	140
117	A genome-wide association study in multiple system atrophy. Neurology, 2016, 87, 1591-1598.	1.5	139
118	Clinical and genetic characterization of families with triple A (Allgrove) syndrome. Brain, 2002, 125, 2681-2690.	3.7	137
119	Unusual phenotypes in DYT1 dystonia: A report of five cases and a review of the literature. Movement Disorders, 2003, 18, 706-711.	2.2	137
120	Calcium is a key factor in Î±-synuclein induced neurotoxicity. Journal of Cell Science, 2016, 129, 1792-801.	1.2	136
121	Cytochrome c Oxidase Deficiency Associated with the First Stop-Codon Point Mutation in Human mtDNA. American Journal of Human Genetics, 1998, 63, 29-36.	2.6	135
122	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid-? concentrations. Annals of Neurology, 2000, 48, 806-808.	2.8	135
123	Genetic overlap between Alzheimerâ€™s disease and Parkinsonâ€™s disease at the MAPT locus. Molecular Psychiatry, 2015, 20, 1588-1595.	4.1	133
124	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. Brain, 2010, 133, 2136-2147.	3.7	132
125	Signalling properties of inorganic polyphosphate in the mammalian brain. Nature Communications, 2013, 4, 1362.	5.8	132
126	The mitochondrial DNA G13513A transition in ND5 is associated with a LHON/MELAS overlap syndrome and may be a frequent cause of MELAS. Annals of Neurology, 1999, 46, 916-919.	2.8	129

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127	Vascular disease and vascular risk factors in relation to motor features and cognition in early Parkinson's disease. Movement Disorders, 2016, 31, 1518-1526.	2.2	128
128	Regional Distribution of Amyloid-Bri Deposition and Its Association with Neurofibrillary Degeneration in Familial British Dementia. American Journal of Pathology, 2001, 158, 515-526.	1.9	127
129	Chorea-Acanthocytosis: Genetic Linkage to Chromosome 9q21. American Journal of Human Genetics, 1997, 61, 899-908.	2.6	126
130	PINK1 function in health and disease. EMBO Molecular Medicine, 2009, 1, 152-165.	3.3	125
131	Genes and susceptibility to multiple sclerosis. Acta Neurologica Scandinavica, 1995, 91, 43-51.	1.0	124
132	Clinical genetics of familial progressive supranuclear palsy. Brain, 1999, 122, 1233-1245.	3.7	124
133	UCHL-1is not a Parkinson's disease susceptibility gene. Annals of Neurology, 2006, 59, 627-633.	2.8	123
134	Autosomal recessive, DYT2-like primary torsion dystonia. Neurology, 2003, 61, 1801-1803.	1.5	122
135	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
136	Association of slow acetylator genotype for N-acetyltransferase 2 with familial Parkinson's disease. Lancet, The, 1997, 350, 1136-1139.	6.3	121
137	The glucocerobrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. Movement Disorders, 2013, 28, 232-236.	2.2	121
138	Low frequency of pathogenic mutations in the ubiquitin carboxyterminal hydrolase gene in familial ParkinsonÊ¼s disease. NeuroReport, 1999, 10, 427-429.	0.6	119
139	The structure of the tau haplotype in controls and in progressive supranuclear palsy. Human Molecular Genetics, 2004, 13, 1267-1274.	1.4	119
140	DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.13-36.32 in an Italian family with cranial-cervical or upper limb onset. Annals of Neurology, 2001, 49, 362-366.	2.8	118
141	Developing and validating Parkinsonâ€™s disease subtypes and their motor and cognitive progression. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1279-1287.	0.9	116
142	<scp>LRRK</scp> 2 activation controls the repair of damaged endomembranes in macrophages. EMBO Journal, 2020, 39, e104494.	3.5	116
143	An mtDNA Mutation in the Initiation Codon of the Cytochrome C Oxidase Subunit II Gene Results in Lower Levels of the Protein and a Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 1999, 64, 1330-1339.	2.6	115
144	Polygenic risk of <scp>P</scp>arkinson disease is correlated with disease age at onset. Annals of Neurology, 2015, 77, 582-591.	2.8	115

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145	A Novel Prion Disease Associated with Diarrhea and Autonomic Neuropathy. New England Journal of Medicine, 2013, 369, 1904-1914.	13.9	113
146	Tau gene and Parkinson's disease: a case-control study and meta-analysis. Journal of Neurology, Neurosurgery and Psychiatry, 2004, 75, 962-965.	0.9	112
147	A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. Annals of Neurology, 2001, 49, 521-525.	2.8	110
148	Large, expanded repeats in SCA8 are not confined to patients with cerebellar ataxia. Nature Genetics, 2000, 24, 214-215.	9.4	109
149	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
150	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532.	2.2	108
151	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
152	Dopa-responsive dystonia in British patients: new mutations of the GTP- cyclohydrolase I gene and evidence for genetic heterogeneity. Human Molecular Genetics, 1996, 5, 403-406.	1.4	107
153	Identifying Candidate Causal Variants Responsible for Altered Activity of the ABCB1 Multidrug Resistance Gene. Genome Research, 2004, 14, 1333-1344.	2.4	107
154	Origin of the Mutations in the parkin Gene in Europe: Exon Rearrangements Are Independent Recurrent Events, whereas Point Mutations May Result from Founder Effects. American Journal of Human Genetics, 2001, 68, 617-626.	2.6	106
155	Normal variation in fronto-occipital circuitry and cerebellar structure with an autism-associated polymorphism of CNTNAP2. NeuroImage, 2010, 53, 1030-1042.	2.1	105
156	Striatal and cortical pre- and postsynaptic dopaminergic dysfunction in sporadic parkin-linked parkinsonism. Brain, 2004, 127, 1332-1342.	3.7	104
157	Nova2 Interacts with a Cis-Acting Polymorphism to Influence the Proportions of Drug-Responsive Splice Variants of SCN1A. American Journal of Human Genetics, 2007, 80, 876-883.	2.6	104
158	Features of GBA-associated Parkinsonâ€™s disease at presentation in the UK Tracking Parkinsonâ€™s study. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 702-709.	0.9	103
159	The genetics of dystonia: new twists in an old tale. Brain, 2013, 136, 2017-2037.	3.7	102
160	Genomeâ€Wide Association Studies of Cognitive and Motor Progression in Parkinson's Disease. Movement Disorders, 2021, 36, 424-433.	2.2	101
161	Single-Molecule Imaging of Individual Amyloid Protein Aggregates in Human Biofluids. ACS Chemical Neuroscience, 2016, 7, 399-406.	1.7	99
162	Sequencing of the alpha-synuclein gene in a large series of cases of familial Parkinson's disease fails to reveal any further mutations. The European Consortium on Genetic Susceptibility in Parkinson's Disease (GSPD). Human Molecular Genetics, 1998, 7, 751-753.	1.4	98

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163	Park6-linked parkinsonism occurs in several european families. Annals of Neurology, 2002, 51, 14-18.	2.8	98
164	De Novo Expansion of Intermediate Alleles in Spinocerebellar Ataxia 7. Human Molecular Genetics, 1998, 7, 1809-1813.	1.4	96
165	Molecular and Clinical Study of 18 Families with ADCA Type II: Evidence for Genetic Heterogeneity and De Novo Mutation. American Journal of Human Genetics, 1999, 64, 1594-1603.	2.6	96
166	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
167	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	2.6	96
168	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
169	Somatic and germline mosaicism in sporadic early-onset Alzheimer's disease. Human Molecular Genetics, 2004, 13, 1219-1224.	1.4	93
170	In vivo skeletal muscle mitochondrial function in Leber's hereditary optic neuropathy assessed by31P magnetic resonance spectroscopy. Annals of Neurology, 1997, 42, 573-579.	2.8	91
171	The ?-synuclein Ala53Thr mutation is not a common cause of familial Parkinson's disease: A study of 230 European cases. Annals of Neurology, 1998, 44, 270-273.	2.8	91
172	Genome-wide association studies: the key to unlocking neurodegeneration?. Nature Neuroscience, 2010, 13, 789-794.	7.1	90
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