

Nicholas W. Wood

List of Publications by Year in descending order

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Version: 2024-02-01

472
papers

68,214
citations

729

120
h-index

906

241
g-index

499
all docs

499
docs citations

499
times ranked

57134
citing authors

#	ARTICLE	IF	CITATIONS
1	Dissecting the Phenotype and Genotype of <i>PLA2G6</i> -Related Parkinsonism. <i>Movement Disorders</i> , 2022, 37, 148-161.	2.2	32
2	Cerebellar and Midbrain Lysosomal Enzyme Deficiency in Isolated Dystonia. <i>Movement Disorders</i> , 2022, 37, 875-877.	2.2	1
3	Combining biomarkers for prognostic modelling of Parkinson's disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 707-715.	0.9	9
4	The role of body fat in multiple sclerosis susceptibility and severity: A Mendelian randomisation study. <i>Multiple Sclerosis Journal</i> , 2022, 28, 1673-1684.	1.4	3
5	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. <i>Movement Disorders</i> , 2021, 36, 251-255.	2.2	23
6	Genome-Wide Association Studies of Cognitive and Motor Progression in Parkinson's Disease. <i>Movement Disorders</i> , 2021, 36, 424-433.	2.2	101
7	Analysis of DNMT3 and VAMP4 as genetic modifiers of LRRK2 Parkinson's disease. <i>Neurobiology of Aging</i> , 2021, 97, 148.e17-148.e24.	1.5	16
8	<i>NOTCH2NLC</i> Intermediate-Length Repeat Expansion and Parkinson's Disease in Patients of European Descent. <i>Annals of Neurology</i> , 2021, 89, 633-635.	2.8	7
9	Expanding the Spectrum of <i>AP5Z1</i> -Related Hereditary Spastic Paraplegia (<i>HSPSPG48</i>): A Multicenter Study on a Rare Disease. <i>Movement Disorders</i> , 2021, 36, 1034-1038.	2.2	9
10	<i>RFC1</i> -related ataxia is a mimic of early multiple system atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 444-446.	0.9	25
11	Mitochondrial <i>DNA</i> Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. <i>Annals of Neurology</i> , 2021, 89, 1240-1247.	2.8	12
12	Biallelic variants in <i>TSPOAP1</i> , encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	18
13	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021, 78, 464.	4.5	95
14	Mendelian Randomisation Finds No Causal Association between Urate and Parkinson's Disease Progression. <i>Movement Disorders</i> , 2021, 36, 2182-2187.	2.2	7
15	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021, 90, 35-42.	2.8	29
16	Childhood-Onset Chorea Caused by a Recurrent De Novo <i>DRD2</i> Variant. <i>Movement Disorders</i> , 2021, 36, 1472-1473.	2.2	6
17	Identification of sixteen novel candidate genes for late onset Parkinson's disease. <i>Molecular Neurodegeneration</i> , 2021, 16, 35.	4.4	41
18	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. <i>Annals of Neurology</i> , 2021, 90, 193-202.	2.8	31

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19	Spastic paraplegia preceding PSEN1 -related familial Alzheimer's disease. <i>Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring</i> , 2021, 13, e12186.	1.2	7
20	Mendelian Randomization Studies: A Path to Better Understand Sex and Gender Differences in Parkinson's Disease?. <i>Movement Disorders</i> , 2021, 36, 2220-2222.	2.2	2
21	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. <i>New England Journal of Medicine</i> , 2021, 385, 1868-1880.	13.9	352
22	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	9.4	223
23	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome. <i>Nature Communications</i> , 2021, 12, 7342.	5.8	44
24	Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. <i>Brain</i> , 2020, 143, 234-248.	3.7	149
25	Neuronal intranuclear inclusion disease is genetically heterogeneous. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1716-1725.	1.7	38
26	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. <i>Genetics in Medicine</i> , 2020, 22, 1851-1862.	1.1	30
27	Reply to: "Dopa-Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansions". <i>Movement Disorders</i> , 2020, 35, 1890-1891.	2.2	2
28	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. <i>Brain</i> , 2020, 143, 2771-2787.	3.7	50
29	Using Mendelian randomization to understand and develop treatments for neurodegenerative disease. <i>Brain Communications</i> , 2020, 2, fcaa031.	1.5	12
30	Fibrillation and molecular characteristics are coherent with clinical and pathological features of 4-repeat tauopathy caused by MAPT variant G273R. <i>Neurobiology of Disease</i> , 2020, 146, 105079.	2.1	4
31	Loss of Function Variants in HOPS Complex Genes VPS16 and VPS41 Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , 2020, 88, 867-877.	2.8	70
32	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. <i>European Journal of Human Genetics</i> , 2020, 28, 1763-1768.	1.4	9
33	Genetic and Clinical Heterogeneity in Thirteen New Cases with Aceruloplasminemia. Atypical Anemia as a Clue for an Early Diagnosis. <i>International Journal of Molecular Sciences</i> , 2020, 21, 2374.	1.8	25
34	MYORG-related disease is associated with central pontine calcifications and atypical parkinsonism. <i>Neurology: Genetics</i> , 2020, 6, e399.	0.9	13
35	GGC Repeat Expansion in NOTCH2NLC Is Rare in European Leukoencephalopathy. <i>Annals of Neurology</i> , 2020, 88, 641-642.	2.8	14
36	GGC repeat expansion in NOTCH2NLC is rare in European patients with essential tremor. <i>Brain</i> , 2020, 143, e57-e57.	3.7	13

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37	The repeat variant in MSH3 is not a genetic modifier for spinocerebellar ataxia type 3 and Friedreich's ataxia. <i>Brain</i> , 2020, 143, e25-e25.	3.7	3
38	The influence of microsatellite polymorphisms in sex steroid receptor genes ESR1, ESR2 and AR on sex differences in brain structure. <i>NeuroImage</i> , 2020, 221, 117087.	2.1	7
39	Penetrance of Parkinson's Disease in <i>LRRK2</i> p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , 2020, 35, 774-780.	2.2	57
40	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 901-911.	1.5	78
41	<i>RFC1</i> Intronic Repeat Expansions Absent in Pathologically Confirmed Multiple Systems Atrophy. <i>Movement Disorders</i> , 2020, 35, 1277-1279.	2.2	26
42	<i>LRRK</i> 2 activation controls the repair of damaged endomembranes in macrophages. <i>EMBO Journal</i> , 2020, 39, e104494.	3.5	116
43	Genetic analysis of Mendelian mutations in a large UK population-based Parkinson's disease study. <i>Brain</i> , 2019, 142, 2828-2844.	3.7	62
44	The Parkinson's Disease Mendelian Randomization Research Portal. <i>Movement Disorders</i> , 2019, 34, 1864-1872.	2.2	50
45	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet Neurology</i> , The, 2019, 18, 1091-1102.	4.9	1,414
46	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019, 34, 1851-1863.	2.2	47
47	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019, 34, 460-468.	2.2	66
48	Using global team science to identify genetic parkinson's disease worldwide. <i>Annals of Neurology</i> , 2019, 86, 153-157.	2.8	26
49	<i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5-phosphate supplementation. <i>Annals of Neurology</i> , 2019, 86, 225-240.	2.8	54
50	L-dopa responsiveness in early Parkinson's disease is associated with the rate of motor progression. <i>Parkinsonism and Related Disorders</i> , 2019, 65, 55-61.	1.1	14
51	Delineating the phenotype of autosomal recessive HPCA mutations: Not only isolated dystonia!. <i>Movement Disorders</i> , 2019, 34, 589-592.	2.2	10
52	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2019, 180, 223-231.	1.1	2
53	<i>GLS</i> loss of function causes autosomal recessive spastic ataxia and optic atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 216-221.	1.7	13
54	Genotype-phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. <i>Movement Disorders</i> , 2018, 33, 1119-1129.	2.2	26

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55	Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1230-1232.	0.9	18
56	Sequencing analysis of the SCA6 CAG expansion excludes an influence of repeat interruptions on disease onset. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1226-1227.	0.9	7
57	Features of <i>GBA</i>-associated Parkinsonâ€™s disease at presentation in the UK <i>Tracking Parkinsonâ€™s</i> study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 702-709.	0.9	103
58	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. <i>Neurobiology of Aging</i> , 2018, 64, 159.e5-159.e8.	1.5	30
59	Genetic variation in VAC14 is associated with bacteremia secondary to diverse pathogens in African children. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E3601-E3603.	3.3	12
60	Genome, transcriptome and proteome: the rise of omics data and their integration in biomedical sciences. <i>Briefings in Bioinformatics</i> , 2018, 19, 286-302.	3.2	498
61	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. <i>Frontiers in Cellular Neuroscience</i> , 2018, 12, 429.	1.8	21
62	Developing and validating Parkinsonâ€™s disease subtypes and their motor and cognitive progression. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1279-1287.	0.9	116
63	LRP10 in Î±-synucleinopathies. <i>Lancet Neurology</i> , The, 2018, 17, 1032.	4.9	15
64	LRP10 in Î±-synucleinopathies. <i>Lancet Neurology</i> , The, 2018, 17, 1033-1034.	4.9	11
65	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. <i>Movement Disorders</i> , 2018, 33, 1961-1965.	2.2	38
66	<sc>DNA</sc> repair in trinucleotide repeat ataxias. <i>FEBS Journal</i> , 2018, 285, 3669-3682.	2.2	12
67	Stratification of candidate genes for Parkinsonâ€™s disease using weighted protein-protein interaction network analysis. <i>BMC Genomics</i> , 2018, 19, 452.	1.2	35
68	Frequency of Loss of Function Variants in <i>LRRK2</i> in Parkinson Disease. <i>JAMA Neurology</i> , 2018, 75, 1416.	4.5	66
69	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	6.0	1,085
70	Mendelian randomization study shows no causal relationship between circulating urate levels and Parkinson's disease. <i>Annals of Neurology</i> , 2018, 84, 191-199.	2.8	43
71	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. <i>Human Mutation</i> , 2018, 39, 965-969.	1.1	34
72	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. <i>Neurology</i> , 2018, 90, e2059-e2067.	1.5	35

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73	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. <i>Cell</i> , 2018, 173, 1705-1715.e16.	13.5	623
74	PREDICT-PD: An online approach to prospectively identify risk indicators of Parkinson's disease. <i>Movement Disorders</i> , 2017, 32, 219-226.	2.2	59
75	Utility of the new Movement Disorder Society clinical diagnostic criteria for Parkinson's disease applied retrospectively in a large cohort study of recent onset cases. <i>Parkinsonism and Related Disorders</i> , 2017, 40, 40-46.	1.1	15
76	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017, 57, 247.e9-247.e13.	1.5	108
77	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. <i>Parkinsonism and Related Disorders</i> , 2017, 41, 37-43.	1.1	67
78	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. <i>JAMA Neurology</i> , 2017, 74, 780.	4.5	245
79	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. <i>American Journal of Human Genetics</i> , 2017, 100, 969-977.	2.6	38
80	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017, 49, 223-237.	9.4	186
81	Establishing the role of rare coding variants in known Parkinson's disease risk loci. <i>Neurobiology of Aging</i> , 2017, 59, 220.e11-220.e18.	1.5	15
82	Autonomic Dysfunction in Early Parkinson's Disease: Results from the United Kingdom Tracking Parkinson's Study. <i>Movement Disorders Clinical Practice</i> , 2017, 4, 509-516.	0.8	35
83	Nonsyndromic Parkinson disease in a family with autosomal dominant optic atrophy due to <i>OPA1</i> mutations. <i>Neurology: Genetics</i> , 2017, 3, e188.	0.9	27
84	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017, 140, 3191-3203.	3.7	323
85	Truncating mutations in <i>SPAST</i> patients are associated with a high rate of psychiatric comorbidities in hereditary spastic paraplegia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 681-687.	0.9	30
86	Estimating the causal influence of body mass index on risk of Parkinson disease: A Mendelian randomisation study. <i>PLoS Medicine</i> , 2017, 14, e1002314.	3.9	152
87	Variation in Recent Onset Parkinson's Disease: Implications for Prodromal Detection. <i>Journal of Parkinson's Disease</i> , 2016, 6, 289-300.	1.5	21
88	Statins are underused in recent-onset Parkinson's disease with increased vascular risk: findings from the UK Tracking Parkinson's and Oxford Parkinson's Disease Centre (OPDC) discovery cohorts. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 1183-1190.	0.9	24
89	B48...DNA repair pathways as a common genetic mechanism modulating the age at onset in polyglutamine diseases. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A26.1-A26.	0.9	0
90	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. <i>American Journal of Human Genetics</i> , 2016, 98, 763-771.	2.6	96

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91	Triple trouble: a striking new phenotype or competing genes in a family with hereditary spastic paraplegia. <i>Journal of Neurology</i> , 2016, 263, 1232-1233.	1.8	4
92	Heterogeneity in clinical features and disease severity in ataxia-associated SYNE1 mutations. <i>Journal of Neurology</i> , 2016, 263, 1503-1510.	1.8	24
93	Equating scores of the University of Pennsylvania Smell Identification Test and Sniffin' Sticks test in patients with Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2016, 33, 96-101.	1.1	46
94	Late-onset Lafora disease with prominent parkinsonism due to a rare mutation in <i>EPM2A</i> . <i>Neurology: Genetics</i> , 2016, 2, e101.	0.9	16
95	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. <i>Annals of Neurology</i> , 2016, 79, 983-990.	2.8	183
96	A genome-wide association study in multiple system atrophy. <i>Neurology</i> , 2016, 87, 1591-1598.	1.5	139
97	Vascular disease and vascular risk factors in relation to motor features and cognition in early Parkinson's disease. <i>Movement Disorders</i> , 2016, 31, 1518-1526.	2.2	128
98	<i>SLC25A46</i> mutations underlie progressive myoclonic ataxia with optic atrophy and neuropathy. <i>Movement Disorders</i> , 2016, 31, 1249-1251.	2.2	49
99	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	9.4	494
100	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. <i>Brain</i> , 2016, 139, 1904-1918.	3.7	170
101	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. <i>Human Molecular Genetics</i> , 2016, 25, dww348.	1.4	48
102	EFFECTS OF VASCULAR COMORBIDITY IN PARKINSON'S DISEASE. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, e1.13-e1.	0.9	0
103	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. <i>American Journal of Human Genetics</i> , 2016, 98, 1092-1100.	2.6	39
104	Olfaction in <i>Parkin</i> single and compound heterozygotes in a cohort of young onset Parkinson's disease patients. <i>Acta Neurologica Scandinavica</i> , 2016, 134, 271-276.	1.0	21
105	Single-Molecule Imaging of Individual Amyloid Protein Aggregates in Human Biofluids. <i>ACS Chemical Neuroscience</i> , 2016, 7, 399-406.	1.7	99
106	Calcium is a key factor in α -synuclein induced neurotoxicity. <i>Journal of Cell Science</i> , 2016, 129, 1792-801.	1.2	136
107	Is the <i>MC1R</i> variant p.R160W associated with Parkinson's?. <i>Annals of Neurology</i> , 2016, 79, 159-161.	2.8	18
108	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. <i>Lancet Neurology</i> , The, 2016, 15, 585-596.	4.9	77

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109	Kinetic model of the aggregation of alpha-synuclein provides insights into prion-like spreading. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E1206-15.	3.3	181
110	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	2.6	333
111	Polygenic risk of ischemic stroke is associated with cognitive ability. <i>Neurology</i> , 2016, 86, 611-618.	1.5	14
112	Alpha-Synuclein Oligomers Interact with Metal Ions to Induce Oxidative Stress and Neuronal Death in Parkinson's Disease. <i>Antioxidants and Redox Signaling</i> , 2016, 24, 376-391.	2.5	266
113	The genetic associations of acute anterior uveitis and their overlap with the genetics of ankylosing spondylitis. <i>Genes and Immunity</i> , 2016, 17, 46-51.	2.2	33
114	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. <i>PLoS Medicine</i> , 2016, 13, e1001976.	3.9	150
115	Ca ²⁺ is a key factor in α -synuclein-induced neurotoxicity. <i>Development (Cambridge)</i> , 2016, 143, e1.1-e1.1.	1.2	5
116	Tracking Parkinson's: Study Design and Baseline Patient Data. <i>Journal of Parkinson's Disease</i> , 2015, 5, 947-959.	1.5	64
117	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 938-947.	2.6	109
118	CHCHD2 and Parkinson's disease. <i>Lancet Neurology</i> , The, 2015, 14, 678-679.	4.9	50
119	Diagnostic clues and manifesting carriers in fukutin-related protein (FKRP) limb-girdle muscular dystrophy. <i>Journal of the Neurological Sciences</i> , 2015, 348, 266-268.	0.3	6
120	Genetic overlap between Alzheimer's disease and Parkinson's disease at the MAPT locus. <i>Molecular Psychiatry</i> , 2015, 20, 1588-1595.	4.1	133
121	Loss-of-function mutations in <i>RAB39B</i> are associated with typical early-onset Parkinson disease. <i>Neurology: Genetics</i> , 2015, 1, e9.	0.9	90
122	Influence of COMT genotype and affective distractors on the processing of self-generated thought. <i>Social Cognitive and Affective Neuroscience</i> , 2015, 10, 777-782.	1.5	11
123	Reply: Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , 2015, 138, e352-e352.	3.7	4
124	<i>ADCY5</i> mutations are another cause of benign hereditary chorea. <i>Neurology</i> , 2015, 85, 80-88.	1.5	140
125	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2015, 36, 1605.e7-1605.e12.	1.5	96
126	Polygenic risk of Parkinson disease is correlated with disease age at onset. <i>Annals of Neurology</i> , 2015, 77, 582-591.	2.8	115

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127	Mutations in HPCA Cause Autosomal-Recessive Primary Isolated Dystonia. <i>American Journal of Human Genetics</i> , 2015, 96, 657-665.	2.6	151
128	Structural characterization of toxic oligomers that are kinetically trapped during α -synuclein fibril formation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, E1994-2003.	3.3	384
129	The <i>CACNA1B</i> R1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. <i>Human Molecular Genetics</i> , 2015, 24, 5326-5329.	1.4	28
130	Preliminary investigation of the influence of dopamine regulating genes on social working memory. <i>Social Neuroscience</i> , 2014, 9, 437-451.	0.7	14
131	Mutations in <i>SNX14</i> Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 611-621.	2.6	89
132	<i>PINK1</i> deficiency in β -cells increases basal insulin secretion and improves glucose tolerance in mice. <i>Open Biology</i> , 2014, 4, 140051.	1.5	40
133	<i>ALS2</i> mutations. <i>Neurology</i> , 2014, 82, 1065-1067.	1.5	29
134	The phenotypic spectrum of <i>DYT24</i> due to <i>ANO3</i> mutations. <i>Movement Disorders</i> , 2014, 29, 928-934.	2.2	161
135	Hypersomnia with dilated pupils in adenosine monophosphate deaminase (<i>AMPD</i>) deficiency. <i>Journal of Sleep Research</i> , 2014, 23, 118-120.	1.7	2
136	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , 2014, 23, 562-562.	1.4	5
137	Autosomal-recessive cerebellar ataxia caused by a novel <i>ADCK3</i> mutation that elongates the protein: clinical, genetic and biochemical characterisation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 493-498.	0.9	48
138	Susceptibility loci for pigmentation and melanoma in relation to Parkinson's disease. <i>Neurobiology of Aging</i> , 2014, 35, 1512.e5-1512.e10.	1.5	28
139	Advances in the Genetics of Parkinson's Disease: A Guide for the Clinician. <i>Movement Disorders Clinical Practice</i> , 2014, 1, 3-13.	0.8	17
140	Rare Individual Amyloid- β Oligomers Act on Astrocytes to Initiate Neuronal Damage. <i>Biochemistry</i> , 2014, 53, 2442-2453.	1.2	83
141	Analysis of Parkinson's disease brain-derived DNA for α -synuclein coding somatic mutations. <i>Movement Disorders</i> , 2014, 29, 1060-1064.	2.2	22
142	Genetic comorbidities in Parkinson's disease. <i>Human Molecular Genetics</i> , 2014, 23, 831-841.	1.4	57
143	When the penny drops. <i>Practical Neurology</i> , 2014, 14, 409-414.	0.5	0
144	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , 2014, 83, 1873-1875.	1.5	33

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145	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. <i>Nature Genetics</i> , 2014, 46, 989-993.	9.4	1,685
146	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , 2014, 137, 2480-2492.	3.7	169
147	Screening of mutations in NOL3 in a myoclonic syndromes series. <i>Journal of Neurology</i> , 2014, 261, 1830-1831.	1.8	1
148	Assessment of Parkinson's disease risk loci in Greece. <i>Neurobiology of Aging</i> , 2014, 35, 442.e9-442.e16.	1.5	18
149	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. <i>Nature Communications</i> , 2014, 5, 4204.	5.8	72
150	Alpha-Synuclein Modulates [Ca ²⁺] _i of Neurons and Astrocytes that Trigger Cell Death. <i>Biophysical Journal</i> , 2014, 106, 529a.	0.2	0
151	Alpha-Synuclein Induces Mitochondrial Dysfunction Leading to a Higher Susceptibility of PTP Opening. <i>Biophysical Journal</i> , 2014, 106, 590a.	0.2	0
152	Friedreich's ataxia and other hereditary ataxias in Greece: An 18-year perspective. <i>Journal of the Neurological Sciences</i> , 2014, 336, 87-92.	0.3	13
153	A Genome-wide Association Analysis of a Broad Psychosis Phenotype Identifies Three Loci for Further Investigation. <i>Biological Psychiatry</i> , 2014, 75, 386-397.	0.7	44
154	No pathogenic <i>GNAL</i> mutations in 192 sporadic and familial cases of cervical dystonia. <i>Movement Disorders</i> , 2014, 29, 154-155.	2.2	8
155	The Parkinson's disease-linked proteins Fbxo7 and Parkin interact to mediate mitophagy. <i>Nature Neuroscience</i> , 2013, 16, 1257-1265.	7.1	292
156	Analysis of Genome-Wide Association Studies of Alzheimer Disease and of Parkinson Disease to Determine If These 2 Diseases Share a Common Genetic Risk. <i>JAMA Neurology</i> , 2013, 70, 1268-76.	4.5	51
157	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. <i>Nature Genetics</i> , 2013, 45, 1150-1159.	9.4	1,395
158	The frequency of spinocerebellar ataxia type 23 in a UK population. <i>Journal of Neurology</i> , 2013, 260, 856-859.	1.8	12
159	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. <i>Journal of Neurology</i> , 2013, 260, 656-660.	1.8	17
160	The glucocerebrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. <i>Movement Disorders</i> , 2013, 28, 232-236.	2.2	121
161	A Novel Prion Disease Associated with Diarrhea and Autonomic Neuropathy. <i>New England Journal of Medicine</i> , 2013, 369, 1904-1914.	13.9	113
162	The Role of the Mitochondrial NCX in the Mechanism of Neurodegeneration in Parkinson's Disease. <i>Advances in Experimental Medicine and Biology</i> , 2013, 961, 241-249.	0.8	25

#	ARTICLE	IF	CITATIONS
163	Common variants in the HLA-DRB1–HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. <i>Nature Genetics</i> , 2013, 45, 208-213.	9.4	86
164	Mutational analysis of <i><scp>PMP22</scp></i>, <i><scp>EGR2</scp></i>, <i><scp>LITAF</scp></i> and <i><scp>NEFL</scp></i> in Greek Charcot–Marie–Tooth type 1 patients. <i>Clinical Genetics</i> , 2013, 83, 388-391.	1.0	2
165	Signalling properties of inorganic polyphosphate in the mammalian brain. <i>Nature Communications</i> , 2013, 4, 1362.	5.8	132
166	The genetics of dystonia: new twists in an old tale. <i>Brain</i> , 2013, 136, 2017-2037.	3.7	102
167	Ataxia telangiectasia presenting as dopa-responsive cervical dystonia. <i>Neurology</i> , 2013, 81, 1148-1151.	1.5	65
168	The Role of Interruptions in polyQ in the Pathology of SCA1. <i>PLoS Genetics</i> , 2013, 9, e1003648.	1.5	73
169	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2013, 22, 1696-1696.	1.4	3
170	Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus. <i>Human Molecular Genetics</i> , 2013, 22, 4653-4660.	1.4	29
171	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , 2013, 22, 1039-1049.	1.4	122
172	FAMILY HISTORY IN YOUNG ONSET PARKINSON'S DISEASE. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, e2.69-e2.	0.9	0
173	<i><scp>C9ORF72</scp></i> expansions, parkinsonism, and Parkinson disease. <i>Neurology</i> , 2013, 81, 808-811.	1.5	57
174	Mutations in the autoregulatory domain of β -tubulin 4a cause hereditary dystonia. <i>Annals of Neurology</i> , 2013, 73, 546-553.	2.8	148
175	TRACKING PARKINSON'S (THE PROBAND STUDY)–INTERIM REPORT FROM THE FIRST 1000 CASES. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2013, 84, e2.70-e2.	0.9	0
176	Genotype and phenotype in Parkinson's disease: Lessons in heterogeneity from deep brain stimulation. <i>Movement Disorders</i> , 2013, 28, 1370-1375.	2.2	77
177	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. <i>PLoS ONE</i> , 2013, 8, e70724.	1.1	45
178	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2012, 21, 4996-5009.	1.4	176
179	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. <i>PLoS Genetics</i> , 2012, 8, e1002548.	1.5	495
180	Myoclonus-dystonia syndrome due to tyrosine hydroxylase deficiency. <i>Neurology</i> , 2012, 79, 435-441.	1.5	45

#	ARTICLE	IF	CITATIONS
181	Dopamine Induced Neurodegeneration in a PINK1 Model of Parkinson's Disease. PLoS ONE, 2012, 7, e37564.	1.1	66
182	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
183	<i>PRRT2</i> gene mutations. Neurology, 2012, 79, 2115-2121.	1.5	159
184	HtrA2 deficiency causes mitochondrial uncoupling through the F1F0-ATP synthase and consequent ATP depletion. Cell Death and Disease, 2012, 3, e335-e335.	2.7	32
185	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
186	Phosphorylation of HtrA2 by cyclin-dependent kinase-5 is important for mitochondrial function. Cell Death and Differentiation, 2012, 19, 257-266.	5.0	35
187	Novel peripheral myelin protein 22 (PMP22) micromutations associated with variable phenotypes in Greek patients with Charcot-Marie-Tooth disease. Brain, 2012, 135, e217-e217.	3.7	9
188	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. Neurology, 2012, 79, 127-131.	1.5	35
189	Use of support vector machines for disease risk prediction in genome-wide association studies: Concerns and opportunities. Human Mutation, 2012, 33, 1708-1718.	1.1	42
190	Young-onset parkinsonism due to homozygous duplication of α -synuclein in a consanguineous family. Movement Disorders, 2012, 27, 1829-1830.	2.2	27
191	Genetic screening of Greek patients with Huntington's disease phenocopies identifies an SCA8 expansion. Journal of Neurology, 2012, 259, 1874-1878.	1.8	20
192	Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke. Nature Genetics, 2012, 44, 328-333.	9.4	375
193	Screening for VPS35 mutations in Parkinson's disease. Neurobiology of Aging, 2012, 33, 838.e1-838.e5.	1.5	53
194	Tau acts as an independent genetic risk factor in pathologically proven PD. Neurobiology of Aging, 2012, 33, 838.e7-838.e11.	1.5	23
195	Study of the genetic variability in a Parkinson's Disease gene: EIF4G1. Neuroscience Letters, 2012, 518, 19-22.	1.0	35
196	Direct Observation of the Interconversion of Normal and Toxic Forms of α -Synuclein. Cell, 2012, 149, 1048-1059.	13.5	755
197	Mutations in the Gene PRRT2 Cause Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions. Cell Reports, 2012, 1, 2-12.	2.9	250
198	Glucocerebrosidase deficiency in substantia nigra of parkinson disease brains. Annals of Neurology, 2012, 72, 455-463.	2.8	473

#	ARTICLE	IF	CITATIONS
199	Systematic Review and UK-Based Study of <i>PARK2</i> (parkin), <i>PINK1</i> , <i>PARK7</i> (DJ-1) and <i>LRRK2</i> in early-onset Parkinson's disease. <i>Movement Disorders</i> , 2012, 27, 1522-1529.	2.2	141
200	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. <i>Nature Genetics</i> , 2012, 44, 1341-1348.	9.4	848
201	Mutations in <i>ANO3</i> Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. <i>American Journal of Human Genetics</i> , 2012, 91, 1041-1050.	2.6	224
202	Genome-Wide Association Study Implicates HLA-C*01:02 as a Risk Factor at the Major Histocompatibility Complex Locus in Schizophrenia. <i>Biological Psychiatry</i> , 2012, 72, 620-628.	0.7	156
203	Analysis of spinocerebellar ataxias due to expanded triplet repeats in Greek patients with cerebellar ataxia. <i>Journal of the Neurological Sciences</i> , 2012, 318, 178-180.	0.3	7
204	Characterisation and Validation of Insertions and Deletions in 173 Patient Exomes. <i>PLoS ONE</i> , 2012, 7, e51292.	1.1	8
205	Genetic linkage analysis of a large family with photoparoxysmal response. <i>Epilepsy Research</i> , 2012, 99, 38-45.	0.8	1
206	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. <i>Movement Disorders</i> , 2012, 27, 526-532.	2.2	108
207	Cooperative Genome-Wide Analysis Shows Increased Homozygosity in Early Onset Parkinson's Disease. <i>PLoS ONE</i> , 2012, 7, e28787.	1.1	21
208	Mutations in Nuclear Genes That Affect Mitochondrial Function in Parkinson's Disease. , 2012, , 43-61.		0
209	Ataxia in a young patient. <i>Practical Neurology</i> , 2011, 11, 319-322.	0.5	0
210	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. <i>Nature</i> , 2011, 476, 214-219.	13.7	2,400
211	Detection of novel mutations and review of published data suggests that hereditary spastic paraplegia caused by spastin (SPAST) mutations is found more often in males. <i>Journal of the Neurological Sciences</i> , 2011, 306, 62-65.	0.3	31
212	Mutational analysis of parkin and PINK1 in multiple system atrophy. <i>Neurobiology of Aging</i> , 2011, 32, 548.e5-548.e7.	1.5	16
213	Bioenergetic Consequences of PINK1 Mutations in Parkinson Disease. <i>PLoS ONE</i> , 2011, 6, e25622.	1.1	88
214	Interview: The genetics of Parkinson's disease: piecing together the jigsaw. <i>Neurodegenerative Disease Management</i> , 2011, 1, 105-107.	1.2	0
215	<i>LRRK2</i> expression in idiopathic and G2019S positive Parkinson's disease subjects: a morphological and quantitative study. <i>Neuropathology and Applied Neurobiology</i> , 2011, 37, 777-790.	1.8	44
216	Common variants near <i>ATM</i> are associated with glycemic response to metformin in type 2 diabetes. <i>Nature Genetics</i> , 2011, 43, 117-120.	9.4	390

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217	Parkinson's disease and cancer: two wars, one front. <i>Nature Reviews Cancer</i> , 2011, 11, 813-823.	12.8	146
218	Mitophagy and Parkinson's disease: The PINK1- <i>parkin</i> link. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2011, 1813, 623-633.	1.9	176
219	Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet, The</i> , 2011, 377, 641-649.	6.3	845
220	Interaction between ERAP1 and HLA-B27 in ankylosing spondylitis implicates peptide handling in the mechanism for HLA-B27 in disease susceptibility. <i>Nature Genetics</i> , 2011, 43, 761-767.	9.4	778
221	An intragenic duplication in guanosine triphosphate cyclohydrolase-1 gene in a dopa-responsive dystonia family. <i>Movement Disorders</i> , 2011, 26, 905-909.	2.2	12
222	Cell metabolism affects selective vulnerability in PINK1-associated Parkinson's disease. <i>Journal of Cell Science</i> , 2011, 124, 4194-4202.	1.2	58
223	Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21. <i>Human Molecular Genetics</i> , 2011, 20, 345-353.	1.4	202
224	PINK1 cleavage at position A103 by the mitochondrial protease PARL. <i>Human Molecular Genetics</i> , 2011, 20, 867-879.	1.4	385
225	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. <i>PLoS Genetics</i> , 2011, 7, e1002142.	1.5	247
226	Rare Deletions at 16p13.11 Predispose to a Diverse Spectrum of Sporadic Epilepsy Syndromes. <i>American Journal of Human Genetics</i> , 2010, 86, 707-718.	2.6	231
227	Nonmotor symptoms in <i>Parkin</i> gene-related parkinsonism. <i>Movement Disorders</i> , 2010, 25, 1279-1284.	2.2	31
228	Atypical parkinsonism with apraxia and supranuclear gaze abnormalities in type 1 Gaucher disease. Expanding the spectrum: Case report and literature review. <i>Movement Disorders</i> , 2010, 25, 1506-1509.	2.2	21
229	Effects of age and MAOA genotype on the neural processing of social rejection. <i>Genes, Brain and Behavior</i> , 2010, 9, 628-637.	1.1	35
230	Genetic variability at the PARK16 locus. <i>European Journal of Human Genetics</i> , 2010, 18, 1356-1359.	1.4	85
231	Genetic and functional characterisation of the P/Q calcium channel in episodic ataxia with epilepsy. <i>Journal of Physiology</i> , 2010, 588, 1905-1913.	1.3	85
232	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. <i>Nature Genetics</i> , 2010, 42, 985-990.	9.4	918
233	Genome-wide association studies: the key to unlocking neurodegeneration?. <i>Nature Neuroscience</i> , 2010, 13, 789-794.	7.1	90
234	Endothelial, Sympathetic, and Cardiac Function in Inherited (6- <i>R</i>)- <i>Erythro-5,6,7,8-Tetrahydro-<i>Biopterin</i> Deficiency</i> . <i>Circulation: Cardiovascular Genetics</i> , 2010, 3, 513-522.	5.1	15

#	ARTICLE	IF	CITATIONS
235	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. <i>Brain</i> , 2010, 133, 2136-2147.	3.7	132
236	Cancer and Neurodegeneration: Between the Devil and the Deep Blue Sea. <i>PLoS Genetics</i> , 2010, 6, e1001257.	1.5	141
237	Hyperexcitable Substantia Nigra Dopamine Neurons in <i>PINK1</i> - and <i>HtrA2/Omi</i> -Deficient Mice. <i>Journal of Neurophysiology</i> , 2010, 104, 3009-3020.	0.9	47
238	Targeting mitochondrial dysfunction in neurodegenerative disease: Part II. <i>Expert Opinion on Therapeutic Targets</i> , 2010, 14, 497-511.	1.5	58
239	Normal variation in fronto-occipital circuitry and cerebellar structure with an autism-associated polymorphism of <i>CNTNAP2</i> . <i>NeuroImage</i> , 2010, 53, 1030-1042.	2.1	105
240	Targeting mitochondrial dysfunction in neurodegenerative disease: Part I. <i>Expert Opinion on Therapeutic Targets</i> , 2010, 14, 369-385.	1.5	56
241	In vivo assessment of brain monoamine systems in parkin gene carriers: A PET study. <i>Experimental Neurology</i> , 2010, 222, 120-124.	2.0	25
242	Characterization of <i>PLA2G6</i> as a locus for dystonia-parkinsonism. <i>Annals of Neurology</i> , 2009, 65, 19-23.	2.8	399
243	Genetic Variants of the α -Synuclein Gene <i>SNCA</i> Are Associated with Multiple System Atrophy. <i>PLoS ONE</i> , 2009, 4, e7114.	1.1	144
244	<i>GJB1</i> gene mutations in suspected inflammatory demyelinating neuropathies not responding to treatment. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2009, 80, 699-700.	0.9	29
245	A Genetically Mediated Bias in Decision Making Driven by Failure of Amygdala Control. <i>Journal of Neuroscience</i> , 2009, 29, 5985-5991.	1.7	183
246	Autosomal-dominant <i>GTPCH1</i> -deficient DRD: clinical characteristics and long-term outcome of 34 patients. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2009, 80, 839-845.	0.9	153
247	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. <i>Brain</i> , 2009, 132, 1783-1794.	3.7	612
248	GLUCOCEREBROSIDASE MUTATIONS IN 108 NEUROPATHOLOGICALLY CONFIRMED CASES OF MULTIPLE SYSTEM ATROPHY. <i>Neurology</i> , 2009, 72, 1185-1186.	1.5	60
249	Differential <i>DJ-1</i> gene expression in Parkinson's disease. <i>Neurobiology of Disease</i> , 2009, 36, 393-400.	2.1	42
250	<i>SNCA</i> variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009, 65, 610-614.	2.8	257
251	<i>PINK1</i> function in health and disease. <i>EMBO Molecular Medicine</i> , 2009, 1, 152-165.	3.3	125
252	Evidence for pre and postsynaptic nigrostriatal dysfunction in the fragile X tremor-Ataxia syndrome. <i>Movement Disorders</i> , 2009, 24, 1245-1247.	2.2	15

#	ARTICLE	IF	CITATIONS
253	Nigrostriatal dysfunction in homozygous and heterozygous <i>parkin</i> gene carriers: An ^{18F} dopa PET progression study. <i>Movement Disorders</i> , 2009, 24, 2260-2266.	2.2	44
254	Mitochondrial dysfunction triggered by loss of HtrA2 results in the activation of a brain-specific transcriptional stress response. <i>Cell Death and Differentiation</i> , 2009, 16, 449-464.	5.0	156
255	Transcriptional repression of p53 by parkin and impairment by mutations associated with autosomal recessive juvenile Parkinson's disease. <i>Nature Cell Biology</i> , 2009, 11, 1370-1375.	4.6	173
256	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. <i>Nature Genetics</i> , 2009, 41, 1330-1334.	9.4	483
257	Genome-wide association study reveals genetic risk underlying Parkinson's disease. <i>Nature Genetics</i> , 2009, 41, 1308-1312.	9.4	1,745
258	No major role of common SV2A variation for predisposition or levetiracetam response in epilepsy. <i>Epilepsy Research</i> , 2009, 83, 44-51.	0.8	32
259	Association of MAPT haplotype-tagging SNPs with sporadic Parkinson's disease. <i>Neurobiology of Aging</i> , 2009, 30, 1477-1482.	1.5	48
260	PINK1-Associated Parkinson's Disease Is Caused by Neuronal Vulnerability to Calcium-Induced Cell Death. <i>Molecular Cell</i> , 2009, 33, 627-638.	4.5	584
261	Characterisation of a novel NR4A2 mutation in Parkinson's disease brain. <i>Neuroscience Letters</i> , 2009, 457, 75-79.	1.0	36
262	Cell Death Pathways in Parkinson's Disease: Role of Mitochondria. <i>Antioxidants and Redox Signaling</i> , 2009, 11, 2135-2149.	2.5	70
263	Riluzole treatment, survival and diagnostic criteria in Parkinson plus disorders: The NNIPPS Study. <i>Brain</i> , 2009, 132, 156-171.	3.7	298
264	Molecular basis of Parkinson's disease. <i>NeuroReport</i> , 2009, 20, 150-156.	0.6	69
265	Clinical and genetic analysis of spinocerebellar ataxia type 11. <i>Cerebellum</i> , 2008, 7, 159-164.	1.4	23
266	Cortical α -synuclein load is associated with amyloid- β plaque burden in a subset of Parkinson's disease patients. <i>Acta Neuropathologica</i> , 2008, 115, 417-425.	3.9	146
267	Motor cortical physiology in patients and asymptomatic carriers of parkin gene mutations. <i>Movement Disorders</i> , 2008, 23, 1812-1819.	2.2	29
268	Mitochondrial function and morphology are impaired in <i>parkin</i> mutant fibroblasts. <i>Annals of Neurology</i> , 2008, 64, 555-565.	2.8	339
269	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. <i>Lancet Neurology</i> , The, 2008, 7, 583-590.	4.9	1,340
270	What Have <i>PINK1</i> and <i>HtrA2</i> Genes Told Us about the Role of Mitochondria in Parkinson's Disease?. <i>Annals of the New York Academy of Sciences</i> , 2008, 1147, 30-36.	1.8	22

#	ARTICLE	IF	CITATIONS
271	Clinical heterogeneity and genotype-phenotype correlations in hereditary spastic paraplegia because of Spatacsin mutations (SPG11). <i>European Journal of Neurology</i> , 2008, 15, 1065-1070.	1.7	34
272	Genetic testing in neurology. <i>Medicine</i> , 2008, 36, 566-568.	0.2	0
273	PINK1 Is Necessary for Long Term Survival and Mitochondrial Function in Human Dopaminergic Neurons. <i>PLoS ONE</i> , 2008, 3, e2455.	1.1	273
274	Expression of BRI2 mRNA and protein in normal human brain and familial British dementia: its relevance to the pathogenesis of disease. <i>Neuropathology and Applied Neurobiology</i> , 2008, 34, 492-505.	1.8	28
275	Second consensus statement on the diagnosis of multiple system atrophy. <i>Neurology</i> , 2008, 71, 670-676.	1.5	2,720
276	Genetics of progressive supranuclear palsy. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2008, 89, 475-485.	1.0	0
277	Neuropathology of primary adult-onset dystonia. <i>Neurology</i> , 2008, 70, 695-699.	1.5	42
278	Mutations in the <i>HSP27</i> (<i>HSPB1</i>) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. <i>Neurology</i> , 2008, 71, 1660-1668.	1.5	168
279	Hyposmia in G2019S LRRK2-related parkinsonism. <i>Neurology</i> , 2008, 71, 1021-1026.	1.5	82
280	Multiple mitochondrial DNA deletions in monozygotic twins with OPMD. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2008, 79, 68-71.	0.9	13
281	Clinical and genetic analysis of spinocerebellar ataxia type 11. <i>Cerebellum</i> , 2008, 7, 1-6.	1.4	0
282	Mitochondrial ND5 Gene Variation Associated with Encephalomyopathy and Mitochondrial ATP Consumption. <i>Journal of Biological Chemistry</i> , 2007, 282, 36845-36852.	1.6	59
283	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. <i>PLoS Genetics</i> , 2007, 3, e108.	1.5	269
284	Large-scale pathways-based association study in amyotrophic lateral sclerosis. <i>Brain</i> , 2007, 130, 2292-2301.	3.7	32
285	Targeting amyloid- β^2 in glaucoma treatment. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 13444-13449.	3.3	315
286	Nova2 Interacts with a Cis-Acting Polymorphism to Influence the Proportions of Drug-Responsive Splice Variants of SCN1A. <i>American Journal of Human Genetics</i> , 2007, 80, 876-883.	2.6	104
287	Alternative ion channel splicing in mesial temporal lobe epilepsy and Alzheimer's disease. <i>Genome Biology</i> , 2007, 8, R32.	13.9	38
288	The mitochondrial protease HtrA2 is regulated by Parkinson's disease-associated kinase PINK1. <i>Nature Cell Biology</i> , 2007, 9, 1243-1252.	4.6	441

#	ARTICLE	IF	CITATIONS
289	Mutations in TTBK2, encoding a kinase implicated in tau phosphorylation, segregate with spinocerebellar ataxia type 11. <i>Nature Genetics</i> , 2007, 39, 1434-1436.	9.4	185
290	A Multicenter Study of BRD2 as a Risk Factor for Juvenile Myoclonic Epilepsy. <i>Epilepsia</i> , 2007, 48, 706-712.	2.6	76
291	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. <i>Lancet Neurology</i> , The, 2007, 6, 970-980.	4.9	175
292	Understanding the molecular causes of Parkinson's disease. <i>Trends in Molecular Medicine</i> , 2006, 12, 521-528.	3.5	274
293	Altered cleavage and localization of PINK1 to aggresomes in the presence of proteasomal stress. <i>Journal of Neurochemistry</i> , 2006, 98, 156-169.	2.1	146
294	Expanding insights of mitochondrial dysfunction in Parkinson's disease. <i>Nature Reviews Neuroscience</i> , 2006, 7, 207-219.	4.9	773
295	A pharmacogenetic exploration of vigabatrin-induced visual field constriction. <i>Epilepsy Research</i> , 2006, 70, 144-152.	0.8	26
296	Examining the role of common genetic variation in the $\beta 2$ subunit of the GABAA receptor in epilepsy using tagging SNPs. <i>Epilepsy Research</i> , 2006, 70, 229-238.	0.8	17
297	Nigral degeneration and striatal dopaminergic dysfunction in idiopathic and parkin-linked Parkinson's disease. <i>Movement Disorders</i> , 2006, 21, 299-305.	2.2	18
298	NR4A2 genetic variation in sporadic Parkinson's disease: A genome-wide approach. <i>Movement Disorders</i> , 2006, 21, 1960-1963.	2.2	15
299	The ADH1C stop mutation in multiple system atrophy patients and healthy probands in the United Kingdom and Germany. <i>Movement Disorders</i> , 2006, 21, 2034-2034.	2.2	6
300	UCHL-1 is not a Parkinson's disease susceptibility gene. <i>Annals of Neurology</i> , 2006, 59, 627-633.	2.8	123
301	A heterozygous effect for PINK1 mutations in Parkinson's disease?. <i>Annals of Neurology</i> , 2006, 60, 414-419.	2.8	149
302	Familial dopa-responsive cervical dystonia. <i>Neurology</i> , 2006, 66, 599-601.	1.5	38
303	PINK1 protein in normal human brain and Parkinson's disease. <i>Brain</i> , 2006, 129, 1720-1731.	3.7	291
304	Genetic association studies of complex neurological diseases. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2006, 77, 1302-1304.	0.9	19
305	Mitochondria in Parkinson Disease. <i>Archives of Neurology</i> , 2006, 63, 649.	4.9	27
306	The α -synuclein gene in multiple system atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2006, 77, 464-467.	0.9	45

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307	The syndrome of (predominantly cervical) dystonia and cerebellar ataxia: new cases indicate a distinct but heterogeneous entity. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2006, 78, 774-775.	0.9	19
308	Molecular genetic pathways in Parkinson's disease: a review. <i>Clinical Science</i> , 2005, 109, 355-364.	1.8	37
309	A single-nucleotide polymorphism tagging set for human drug metabolism and transport. <i>Nature Genetics</i> , 2005, 37, 84-89.	9.4	142
310	PARK11 is not linked with Parkinson's disease in European families. <i>European Journal of Human Genetics</i> , 2005, 13, 193-197.	1.4	23
311	Apolipoprotein E4 is probably responsible for the chromosome 19 linkage peak for Parkinson's disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2005, 136B, 72-74.	1.1	27
312	Parkin disease in a Brazilian kindred: Manifesting heterozygotes and clinical follow-up over 10 years. <i>Movement Disorders</i> , 2005, 20, 479-484.	2.2	32
313	Parkinsonism and nigrostriatal dysfunction are associated with spinocerebellar ataxia type 6 (SCA6). <i>Movement Disorders</i> , 2005, 20, 1115-1119.	2.2	45
314	UCHL-1 gene in multiple system atrophy: A haplotype tagging approach. <i>Movement Disorders</i> , 2005, 20, 1338-1343.	2.2	17
315	Commentary on "A genome wide linkage disequilibrium screen in Parkinson's disease". <i>Journal of Neurology</i> , 2005, 252, 603-604.	1.8	0
316	Linkage disequilibrium fine mapping and haplotype association analysis of the tau gene in progressive supranuclear palsy and corticobasal degeneration. <i>Journal of Medical Genetics</i> , 2005, 42, 837-846.	1.5	225
317	Population genetic approaches to neurological disease: Parkinson's disease as an example. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2005, 360, 1573-1578.	1.8	4
318	Introduction: genetic variation and human health. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2005, 360, 1539-1541.	1.8	2
319	Failure to replicate previously reported genetic associations with sporadic temporal lobe epilepsy: where to from here?. <i>Brain</i> , 2005, 128, 1832-1840.	3.7	87
320	Association of genetic loci: Replication or not, that is the question. <i>Neurology</i> , 2005, 64, 1989.	1.5	7
321	Genetic predictors of the maximum doses patients receive during clinical use of the anti-epileptic drugs carbamazepine and phenytoin. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2005, 102, 5507-5512.	3.3	321
322	The sepiapterin reductase gene region reveals association in the PARK3 locus: analysis of familial and sporadic Parkinson's disease in European populations. <i>Journal of Medical Genetics</i> , 2005, 43, 557-562.	1.5	38
323	The fragile X tremor ataxia syndrome in the differential diagnosis of multiple system atrophy: data from the EMSA Study Group. <i>Brain</i> , 2005, 128, 1855-1860.	3.7	172
324	Mutations in the gene LRRK2 encoding dardarin (PARK8) cause familial Parkinson's disease: clinical, pathological, olfactory and functional imaging and genetic data. <i>Brain</i> , 2005, 128, 2786-2796.	3.7	315

#	ARTICLE	IF	CITATIONS
325	A common LRRK2 mutation in idiopathic Parkinson's disease. <i>Lancet, The</i> , 2005, 365, 415-416.	6.3	391
326	Molecular pathogenesis of Parkinson's disease. <i>Human Molecular Genetics</i> , 2005, 14, 2749-2755.	1.4	187
327	Synphilin-1 and parkin show overlapping expression patterns in human brain and form aggregates in response to proteasomal inhibition. <i>Neurobiology of Disease</i> , 2005, 20, 401-411.	2.1	40
328	A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. <i>Brain</i> , 2004, 127, 973-980.	3.7	77
329	The structure of the tau haplotype in controls and in progressive supranuclear palsy. <i>Human Molecular Genetics</i> , 2004, 13, 1267-1274.	1.4	119
330	Genome-wide scan linkage analysis for Parkinson's disease: the European genetic study of Parkinson's disease. <i>Journal of Medical Genetics</i> , 2004, 41, 900-907.	1.5	38
331	Autosomal dominant cerebellar ataxia: SCA2 is the most frequent mutation in eastern India. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2004, 75, 448-452.	0.9	62
332	Tau gene and Parkinson's disease: a case-control study and meta-analysis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2004, 75, 962-965.	0.9	112
333	Genome-Wide Analysis of the Parkinsonism-Dementia Complex of Guam. <i>Archives of Neurology</i> , 2004, 61, 1889-97.	4.9	44
334	The expression of DJ-1 (PARK7) in normal human CNS and idiopathic Parkinson's disease. <i>Brain</i> , 2004, 127, 420-430.	3.7	404
335	Trinucleotide repeats and neurodegenerative disease. <i>Brain</i> , 2004, 127, 2385-2405.	3.7	160
336	The spectrum of pathological involvement of the striatonigral and olivopontocerebellar systems in multiple system atrophy: clinicopathological correlations. <i>Brain</i> , 2004, 127, 2657-2671.	3.7	493
337	Identifying Candidate Causal Variants Responsible for Altered Activity of the ABCB1 Multidrug Resistance Gene. <i>Genome Research</i> , 2004, 14, 1333-1344.	2.4	107
338	Striatal and cortical pre- and postsynaptic dopaminergic dysfunction in sporadic parkin-linked parkinsonism. <i>Brain</i> , 2004, 127, 1332-1342.	3.7	104
339	Assessment of a DJ-1 (PARK7) polymorphism in Finnish PD. <i>Neurology</i> , 2004, 62, 2335-2335.	1.5	2
340	Reduction in endogenous parkin levels renders glial cells sensitive to both caspase-dependent and caspase-independent cell death. <i>European Journal of Neuroscience</i> , 2004, 20, 2038-2048.	1.2	17
341	PINK, PANK, or PARK? A clinicians' guide to familial parkinsonism. <i>Lancet Neurology, The</i> , 2004, 3, 652-662.	4.9	61
342	Genetic causes of Parkinson's disease: UCHL-1. <i>Cell and Tissue Research</i> , 2004, 318, 189-194.	1.5	51

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343	Causes of Parkinson's disease: genetics of DJ-1. <i>Cell and Tissue Research</i> , 2004, 318, 185-188.	1.5	51
344	Genetic Approaches to Solving Common Diseases. <i>Journal of Neurology</i> , 2004, 251, 1169-1172.	1.8	1
345	A functional polymorphism regulating dopamine β -hydroxylase influences against Parkinson's disease. <i>Annals of Neurology</i> , 2004, 55, 443-446.	2.8	59
346	The gene responsible for PARK6 Parkinson's disease, PINK1, does not influence common forms of parkinsonism. <i>Annals of Neurology</i> , 2004, 56, 329-335.	2.8	38
347	Ataxin-7 aggregation and ubiquitination in infantile SCA7 with 180 CAG repeats. <i>Annals of Neurology</i> , 2004, 56, 448-452.	2.8	47
348	Connexin 32 promoter P2 mutations: A mechanism of peripheral nerve dysfunction. <i>Annals of Neurology</i> , 2004, 56, 730-734.	2.8	51
349	Population genetics for target identification. <i>Drug Discovery Today: Technologies</i> , 2004, 1, 69-74.	4.0	10
350	Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. <i>Science</i> , 2004, 304, 1158-1160.	6.0	3,060
351	Cloning of the Gene Containing Mutations that Cause PARK8-Linked Parkinson's Disease. <i>Neuron</i> , 2004, 44, 595-600.	3.8	2,183
352	Somatic and germline mosaicism in sporadic early-onset Alzheimer's disease. <i>Human Molecular Genetics</i> , 2004, 13, 1219-1224.	1.4	93
353	Genome scans and candidate gene approaches in the study of common diseases and variable drug responses. <i>Trends in Genetics</i> , 2003, 19, 615-622.	2.9	151
354	Slowly progressive cerebellar ataxia and cervical dystonia: Clinical presentation of a new form of spinocerebellar ataxia?. <i>Movement Disorders</i> , 2003, 18, 200-206.	2.2	35
355	Unusual phenotypes in DYT1 dystonia: A report of five cases and a review of the literature. <i>Movement Disorders</i> , 2003, 18, 706-711.	2.2	137
356	How much phenotypic variation can be attributed to parkin genotype?. <i>Annals of Neurology</i> , 2003, 54, 176-185.	2.8	271
357	The role of pathogenic DJ-1 mutations in Parkinson's disease. <i>Annals of Neurology</i> , 2003, 54, 283-286.	2.8	362
358	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. <i>Neuropathology and Applied Neurobiology</i> , 2003, 29, 288-302.	1.8	194
359	Association of Multidrug Resistance in Epilepsy with a Polymorphism in the Drug-Transporter Gene ABCB1. <i>New England Journal of Medicine</i> , 2003, 348, 1442-1448.	13.9	690
360	Selection and Evaluation of Tagging SNPs in the Neuronal-Sodium-Channel Gene SCN1A: Implications for Linkage-Disequilibrium Gene Mapping. <i>American Journal of Human Genetics</i> , 2003, 73, 551-565.	2.6	181

#	ARTICLE	IF	CITATIONS
361	Different patterns of electrophysiological deficits in manifesting and non-manifesting carriers of the DYT1 gene mutation. <i>Brain</i> , 2003, 126, 2074-2080.	3.7	141
362	Parkin is recruited into aggresomes in a stress-specific manner: over-expression of parkin reduces aggresome formation but can be dissociated from parkin's effect on neuronal survival. <i>Human Molecular Genetics</i> , 2003, 13, 117-135.	1.4	72
363	Gluten ataxia in perspective: epidemiology, genetic susceptibility and clinical characteristics. <i>Brain</i> , 2003, 126, 685-691.	3.7	248
364	Hereditary sensory neuropathy is caused by a mutation in the delta subunit of the cytosolic chaperonin-containing t-complex peptide-1 (Cct4) gene. <i>Human Molecular Genetics</i> , 2003, 12, 1917-1925.	1.4	51
365	Parkin disease: a phenotypic study of a large case series. <i>Brain</i> , 2003, 126, 1279-1292.	3.7	427
366	Parkin mutations are frequent in patients with isolated early-onset parkinsonism. <i>Brain</i> , 2003, 126, 1271-1278.	3.7	279
367	Autosomal recessive, DYT2-like primary torsion dystonia. <i>Neurology</i> , 2003, 61, 1801-1803.	1.5	122
368	Six novel connexin32 (GJB1) mutations in X-linked Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 73, 304-306.	0.9	56
369	Sequence analysis of tau in familial and sporadic progressive supranuclear palsy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 72, 388-390.	0.9	17
370	Running a neurogenetic clinic. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 73, 2ii-4.	0.9	8
371	Genetics of movement disorders and ataxia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 73, 22ii-26.	0.9	37
372	Neuronal intranuclear inclusions in SCA2: a genetic, morphological and immunohistochemical study of two cases. <i>Brain</i> , 2002, 125, 656-663.	3.7	87
373	Intrafamilial Phenotypic Variability in Friedreich Ataxia Associated With a G130V Mutation in the FRDA Gene. <i>Archives of Neurology</i> , 2002, 59, 296.	4.9	17
374	Clinical and genetic characterization of families with triple A (Allgrove) syndrome. <i>Brain</i> , 2002, 125, 2681-2690.	3.7	137
375	Familial adult onset of Krabbe's disease resembling hereditary spastic paraplegia with normal neuroimaging. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 72, 635-638.	0.9	43
376	Dopa-Responsive Dystonia - The Story so Far. <i>Neuropediatrics</i> , 2002, 33, 1-5.	0.3	60
377	Immunological study of hereditary motor and sensory neuropathy type 1a (HMSN1a). <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2002, 72, 230-235.	0.9	37
378	The tau locus is not significantly associated with pathologically confirmed sporadic Parkinson's disease. <i>Neuroscience Letters</i> , 2002, 330, 201-203.	1.0	39

#	ARTICLE	IF	CITATIONS
379	Progression of nigrostriatal dysfunction in a parkin kindred: an [18F]dopa PET and clinical study. <i>Brain</i> , 2002, 125, 2248-2256.	3.7	141
380	Mouse models for neurological disease. <i>Lancet Neurology</i> , The, 2002, 1, 215-224.	4.9	41
381	Complex relationship between Parkin mutations and Parkinson disease. <i>American Journal of Medical Genetics Part A</i> , 2002, 114, 584-591.	2.4	193
382	Park6-linked parkinsonism occurs in several european families. <i>Annals of Neurology</i> , 2002, 51, 14-18.	2.8	98
383	Partial epilepsy with pericentral spikes: A new familial epilepsy syndrome with evidence for linkage to chromosome 4p15. <i>Annals of Neurology</i> , 2002, 51, 740-749.	2.8	40
384	Myoclonus-dystonia syndrome: $\hat{\mu}$ -sarcoglycan mutations and phenotype. <i>Annals of Neurology</i> , 2002, 52, 489-492.	2.8	143
385	Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: An18F-dopa PET study. <i>Annals of Neurology</i> , 2002, 52, 849-853.	2.8	192
386	Corticobasal degeneration syndrome with basal ganglia calcification: Fahr's disease as a corticobasal look-alike?. <i>Movement Disorders</i> , 2002, 17, 563-567.	2.2	27
387	Genetic and clinical heterogeneity in paroxysmal kinesigenic dyskinesia: Evidence for a third EKD gene. <i>Movement Disorders</i> , 2002, 17, 717-725.	2.2	85
388	PARK6 is a common cause of familial parkinsonism. <i>Neurological Sciences</i> , 2002, 23, s117-s118.	0.9	34
389	Mutation analysis of the sodium/hydrogen exchanger gene (NHE5) in familial paroxysmal kinesigenic dyskinesia. <i>Journal of Neural Transmission</i> , 2002, 109, 1189-1194.	1.4	12
390	Mutation of the sterol 27-hydroxylase gene (CYP27A1) in a Taiwanese family with cerebrotendinous xanthomatosis. <i>Journal of Neurology</i> , 2002, 249, 1311-1312.	1.8	8
391	Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. <i>European Journal of Human Genetics</i> , 2002, 10, 773-781.	1.4	172
392	Regional Distribution of Amyloid-Bri Deposition and Its Association with Neurofibrillary Degeneration in Familial British Dementia. <i>American Journal of Pathology</i> , 2001, 158, 515-526.	1.9	127
393	Origin of the Mutations in the parkin Gene in Europe: Exon Rearrangements Are Independent Recurrent Events, whereas Point Mutations May Result from Founder Effects. <i>American Journal of Human Genetics</i> , 2001, 68, 617-626.	2.6	106
394	Localization of a Novel Locus for Autosomal Recessive Early-Onset Parkinsonism, PARK6, on Human Chromosome 1p35-p36. <i>American Journal of Human Genetics</i> , 2001, 68, 895-900.	2.6	459
395	Localization of the Gene for Distal Hereditary Motor Neuronopathy VII (dHMN-VII) to Chromosome 2q14. <i>American Journal of Human Genetics</i> , 2001, 68, 1270-1276.	2.6	68
396	No pathogenic mutations in the synphilin-1 gene in Parkinson's disease. <i>Neuroscience Letters</i> , 2001, 307, 125-127.	1.0	18

#	ARTICLE	IF	CITATIONS
397	Strong association of a novel Tau promoter haplotype in progressive supranuclear palsy. <i>Neuroscience Letters</i> , 2001, 311, 145-148.	1.0	49
398	Effect of ApoE and tau on age of onset of progressive supranuclear palsy and multiple system atrophy. <i>Neuroscience Letters</i> , 2001, 312, 118-120.	1.0	26
399	Chromosome 13 dementia syndromes as models of neurodegeneration. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2001, 8, 277-284.	1.4	29
400	Identification of a novel primary torsion dystonia locus (DYT13) on chromosome 1p36 in an Italian family with cranial-cervical or upper limb onset. <i>Neurological Sciences</i> , 2001, 22, 95-96.	0.9	23
401	Parkinson's disease is not associated with the combined α -synuclein/apolipoprotein E susceptibility genotype. <i>Annals of Neurology</i> , 2001, 49, 665-668.	2.8	66
402	A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. <i>Annals of Neurology</i> , 2001, 49, 521-525.	2.8	110
403	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. <i>Annals of Neurology</i> , 2001, 49, 362-366.	2.8	118
404	A conserved sorting-associated protein is mutant in chorea-acanthocytosis. <i>Nature Genetics</i> , 2001, 28, 119-120.	9.4	357
405	Systemic Amyloid Deposits in Familial British Dementia. <i>Journal of Biological Chemistry</i> , 2001, 276, 43909-43914.	1.6	73
406	Cervical dystonia is associated with a polymorphism in the dopamine (D5) receptor gene. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2001, 71, 262-264.	0.9	50
407	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. , 2001, 49, 362.		3
408	The paroxysmal dyskinesias. , 2001, , 125-140.		11
409	Genetics of the overlap between epilepsy and movement disorders. , 2001, , 451-464.		0
410	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid- β concentrations. <i>Annals of Neurology</i> , 2000, 48, 806-808.	2.8	135
411	Detailed genotyping demonstrates association between the slow acetylator genotype for N-Acetyltransferase 2 (NAT2) and familial parkinson's disease. <i>Movement Disorders</i> , 2000, 15, 30-35.	2.2	37
412	Paroxysmal dystonic choreoathetosis: Clinical features and investigation of pathophysiology in a large family. <i>Movement Disorders</i> , 2000, 15, 648-657.	2.2	46
413	Large, expanded repeats in SCA8 are not confined to patients with cerebellar ataxia. <i>Nature Genetics</i> , 2000, 24, 214-215.	9.4	109
414	A new family with paroxysmal exercise induced dystonia and migraine: a clinical and genetic study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2000, 68, 609-614.	0.9	72

#	ARTICLE	IF	CITATIONS
415	Mitochondrial DNA Point Mutation T9176C in Leigh Syndrome. <i>Journal of Child Neurology</i> , 2000, 15, 830-833.	0.7	19
416	Association between a polymorphism of ubiquitin carboxy-terminal hydrolase L1 (UCH-L1) gene and sporadic Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2000, 6, 195-197.	1.1	61
417	The genetics of Parkinson's disease. <i>Current Opinion in Genetics and Development</i> , 2000, 10, 292-298.	1.5	73
418	Association between Early-Onset Parkinson's Disease and Mutations in the Parkin Gene. <i>New England Journal of Medicine</i> , 2000, 342, 1560-1567.	13.9	1,448
419	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS1 mutations that lead to exceptionally high amyloid β concentrations. <i>Annals of Neurology</i> , 2000, 48, 806-808.	2.8	3
420	Clinical genetics of familial progressive supranuclear palsy. <i>Brain</i> , 1999, 122, 1233-1245.	3.7	124
421	Huntington's disease progression. <i>Brain</i> , 1999, 122, 2353-2363.	3.7	193
422	A novel mutation in the human voltage-gated potassium channel gene (Kv1.1) associates with episodic ataxia type 1 and sometimes with partial epilepsy. <i>Brain</i> , 1999, 122, 817-825.	3.7	314
423	Phenotypic variation of a new PO mutation in genetically identical twins. <i>Journal of Neurology</i> , 1999, 246, 596-599.	1.8	35
424	The mitochondrial DNA G13513A transition in ND5 is associated with a LHON/MELAS overlap syndrome and may be a frequent cause of MELAS. <i>Annals of Neurology</i> , 1999, 46, 916-919.	2.8	129
425	Neurofibrillary tangle parkinsonian disorders? tau pathology and tau genetics. <i>Movement Disorders</i> , 1999, 14, 731-736.	2.2	40
426	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. <i>American Journal of Human Genetics</i> , 1999, 64, 1330-1339.	2.6	115
427	Molecular and Clinical Study of 18 Families with ADCA Type II: Evidence for Genetic Heterogeneity and De Novo Mutation. <i>American Journal of Human Genetics</i> , 1999, 64, 1594-1603.	2.6	96
428	Autosomal Dominant Cerebellar Ataxia Type III: Linkage in a Large British Family to a 7.6-cM Region on Chromosome 15q14-21.3. <i>American Journal of Human Genetics</i> , 1999, 65, 420-426.	2.6	140
429	The Ile93Met mutation in the ubiquitin carboxy-terminal-hydrolase-L1 gene is not observed in European cases with familial Parkinson's disease. <i>Neuroscience Letters</i> , 1999, 270, 1-4.	1.0	75
430	Mutations in the gene encoding human porsyn are not associated with amyotrophic lateral sclerosis or familial Parkinson's disease. <i>Neuroscience Letters</i> , 1999, 274, 21-24.	1.0	16
431	Prader-Willi and Angelman syndromes: update on genetic mechanisms and diagnostic complexities. <i>Current Opinion in Neurology</i> , 1999, 12, 149-154.	1.8	23
432	Low frequency of pathogenic mutations in the ubiquitin carboxyterminal hydrolase gene in familial Parkinson's disease. <i>NeuroReport</i> , 1999, 10, 427-429.	0.6	119

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433	The mitochondrial DNA G13513A transition in ND5 is associated with a LHON/MELAS overlap syndrome and may be a frequent cause of MELAS. , 1999, 46, 916.		2
434	The genetics of Parkinson's disease. <i>Current Opinion in Neurology</i> , 1999, 12, 427-432.	1.8	15
435	Dejerine-sottas neuropathy and PMP22 point mutations: A new base pair substitution and a possible 'hot spot' on Ser72. <i>Annals of Neurology</i> , 1998, 43, 680-683.	2.8	42
436	The α -synuclein Ala53Thr mutation is not a common cause of familial Parkinson's disease: A study of 230 European cases. <i>Annals of Neurology</i> , 1998, 44, 270-273.	2.8	91
437	Dopa-responsive dystonia: A clinical and molecular genetic study. <i>Annals of Neurology</i> , 1998, 44, 649-656.	2.8	153
438	Genetic aspects of Parkinson's disease. <i>Movement Disorders</i> , 1998, 13, 203-211.	2.2	38
439	Generalized chorea in two patients harboring the Friedreich's ataxia gene trinucleotide repeat expansion. <i>Movement Disorders</i> , 1998, 13, 339-340.	2.2	48
440	Homozygous deletions in parkin gene in European and North African families with autosomal recessive juvenile parkinsonism. <i>Lancet</i> , The, 1998, 352, 1355-1356.	6.3	199
441	Cytochrome c Oxidase Deficiency Associated with the First Stop-Codon Point Mutation in Human mtDNA. <i>American Journal of Human Genetics</i> , 1998, 63, 29-36.	2.6	135
442	De Novo Expansion of Intermediate Alleles in Spinocerebellar Ataxia 7. <i>Human Molecular Genetics</i> , 1998, 7, 1809-1813.	1.4	96
443	Fine mapping of de novo CMT1A and HNPP rearrangements within CMT1A-REPs evidences two distinct sex-dependent mechanisms and candidate sequences involved in recombination. <i>Human Molecular Genetics</i> , 1998, 7, 141-148.	1.4	85
444	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). <i>Human Molecular Genetics</i> , 1998, 7, 751-753.	1.4	98
445	Genetic risk factors in parkinson's disease. <i>Annals of Neurology</i> , 1998, 44, S58-S62.	2.8	15
446	The role of the SCA2 trinucleotide repeat expansion in 89 autosomal dominant cerebellar ataxia families. Frequency, clinical and genetic correlates. <i>Brain</i> , 1998, 121, 459-467.	3.7	84
447	Paroxysmal dystonic choreoathetosis. Genetic linkage studies in a British family. <i>Brain</i> , 1997, 120, 2125-2130.	3.7	39
448	Identification and sizing of the GAA trinucleotide repeat expansion of Friedreich's ataxia in 56 patients. Clinical and genetic correlates. <i>Brain</i> , 1997, 120, 673-680.	3.7	77
449	Association of slow acetylator genotype for N-acetyltransferase 2 with familial Parkinson's disease. <i>Lancet</i> , The, 1997, 350, 1136-1139.	6.3	121
450	Chorea-Acanthocytosis: Genetic Linkage to Chromosome 9q21. <i>American Journal of Human Genetics</i> , 1997, 61, 899-908.	2.6	126

#	ARTICLE	IF	CITATIONS
451	Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion. <i>Nature Genetics</i> , 1997, 17, 65-70.	9.4	758
452	Sex-dependent rearrangements resulting in CMT1A and HNPP. <i>Nature Genetics</i> , 1997, 17, 136-137.	9.4	57
453	Depletion of mitochondrial DNA by ddC in untransformed human cell lines. <i>Somatic Cell and Molecular Genetics</i> , 1997, 23, 287-290.	0.7	45
454	Mitochondrial DNA polymorphisms in pathologically proven Parkinson's disease. <i>Journal of Neurology</i> , 1997, 244, 262-265.	1.8	50
455	In vivo skeletal muscle mitochondrial function in Leber's hereditary optic neuropathy assessed by ³¹ P magnetic resonance spectroscopy. <i>Annals of Neurology</i> , 1997, 42, 573-579.	2.8	91
456	Progressive cognitive decline with truncal/limb ataxia and ballistic movements. <i>Movement Disorders</i> , 1997, 12, 1075-1084.	2.2	0
457	The GTP-cyclohydrolase I gene in atypical Parkinsonian patients: a clinico-genetic study. <i>Journal of the Neurological Sciences</i> , 1996, 141, 27-32.	0.3	23
458	The human homologue of the weaver mouse gene in familial and sporadic Parkinson's disease. <i>Neuroscience</i> , 1996, 72, 877-879.	1.1	23
459	Mitochondrial disorders in neuro-ophthalmology. <i>Current Opinion in Neurology</i> , 1996, 9, 1-4.	1.8	3
460	Dopa-responsive dystonia in British patients: new mutations of the GTP- cyclohydrolase I gene and evidence for genetic heterogeneity. <i>Human Molecular Genetics</i> , 1996, 5, 403-406.	1.4	107
461	Genes and susceptibility to multiple sclerosis. <i>Acta Neurologica Scandinavica</i> , 1995, 91, 43-51.	1.0	124
462	Susceptibility to multiple sclerosis and the immunoglobulin heavy chain variable region. <i>Journal of Neurology</i> , 1995, 242, 677-682.	1.8	36
463	Multiple sclerosis and the HLA-D region: linkage and association studies. <i>Journal of Neuroimmunology</i> , 1995, 58, 183-190.	1.1	64
464	No linkage or association between multiple sclerosis and the myelin basic protein gene in affected sibling pairs. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 1994, 57, 1191-1194.	0.9	33
465	No linkage between multiple sclerosis and the T cell receptor β chain locus. <i>Journal of the Neurological Sciences</i> , 1994, 124, 32-37.	0.3	13
466	The pathogenesis of demyelinating disease. <i>Progress in Neurobiology</i> , 1994, 43, 143-173.	2.8	53
467	Multiple sclerosis in the Cambridge health district of east Anglia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 1992, 55, 877-882.	0.9	50
468	Pharmacogenomics and the Treatment of Neurological Disease. , 0, , 337-346.		0

#	ARTICLE	IF	CITATIONS
469	Cerebellar Ataxias and Related Conditions. , 0, , 629-643.		0
470	The ataxias. , 0, , 52-63.		0
471	The human genome project “ what it really means and where next. , 0, , 1-5.		1
472	Channelopathies. , 0, , 121-135.		0