Nicholas W. Wood

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6941243/publications.pdf

Version: 2024-02-01

472	68,214	867 120	1056 241
papers	citations	h-index	g-index

499	499	499	62592
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Dissecting the Phenotype and Genotype of <scp>PLA2G6</scp>â€Related Parkinsonism. Movement Disorders, 2022, 37, 148-161.	2.2	32
2	Cerebellar and Midbrain Lysosomal Enzyme Deficiency in Isolated Dystonia. Movement Disorders, 2022, 37, 875-877.	2.2	1
3	Combining biomarkers for prognostic modelling of Parkinsonâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 707-715.	0.9	9
4	The role of body fat in multiple sclerosis susceptibility and severity: A Mendelian randomisation study. Multiple Sclerosis Journal, 2022, 28, 1673-1684.	1.4	3
5	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. Movement Disorders, 2021, 36, 251-255.	2.2	23
6	Genomeâ€Wide Association Studies of Cognitive and Motor Progression in Parkinson's Disease. Movement Disorders, 2021, 36, 424-433.	2.2	101
7	Analysis of DNM3 and VAMP4 as genetic modifiers of LRRK2 Parkinsonâ€™s disease. Neurobiology of Aging, 2021, 97, 148.e17-148.e24.	1.5	16
8	<scp>NOTCH2NLC</scp> Intermediateâ€Length Repeat Expansion and Parkinson's Disease in Patients of European Descent. Annals of Neurology, 2021, 89, 633-635.	2.8	7
9	Expanding the Spectrum of <scp>AP5Z1â€</scp>Related Hereditary Spastic Paraplegia (<scp>HSPâ€SPG48</scp>): A Multicenter Study on a Rare Disease. Movement Disorders, 2021, 36, 1034-1038.	2.2	9
10	RFC1-related ataxia is a mimic of early multiple system atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 444-446.	0.9	25
11	Mitochondrial <scp>D</scp><scp>NA</scp> Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. Annals of Neurology, 2021, 89, 1240-1247.	2.8	12
12	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. Journal of Clinical Investigation, 2021, 131, .	3.9	18
13	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
14	Mendelian Randomisation Finds No Causal Association between Urate and Parkinson's Disease Progression. Movement Disorders, 2021, 36, 2182-2187.	2.2	7
15	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. Annals of Neurology, 2021, 90, 35-42.	2.8	29
16	Childhoodâ€Onset Chorea Caused by a Recurrent De Novo DRD2 Variant. Movement Disorders, 2021, 36, 1472-1473.	2.2	6
17	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	4.4	41
18	Genomeâ€Wide Association Study Identifies Risk Loci for Cluster Headache. Annals of Neurology, 2021, 90, 193-202.	2.8	31

#	Article	IF	Citations
19	Spastic paraplegia preceding PSEN1 â€related familial Alzheimer's disease. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12186.	1.2	7
20	Mendelian Randomization Studies: A Path to Better Understand Sex and Gender Differences in Parkinson's Disease?. Movement Disorders, 2021, 36, 2220-2222.	2.2	2
21	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
22	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
23	Finding genetically-supported drug targets for Parkinsonâ€™s disease using Mendelian randomization of the druggable genome. Nature Communications, 2021, 12, 7342.	5.8	44
24	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
25	Neuronal intranuclear inclusion disease is genetically heterogeneous. Annals of Clinical and Translational Neurology, 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. Genetics in Medicine, 2020, 22, 1851-1862.	1.1	30
27	Reply to: â€œDopaâ€Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansionsâ€. Movement Disorders, 2020, 35, 1890-1891.	2.2	2
28	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. Brain, 2020, 143, 2771-2787.	3.7	50
29	Using Mendelian randomization to understand and develop treatments for neurodegenerative disease. Brain Communications, 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. Neurobiology of Disease, 2020, 146, 105079.	2.1	4
31	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	2.8	70
32	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. European Journal of Human Genetics, 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. International Journal of Molecular Sciences, 2020, 21, 2374.	1.8	25
34	MYORG -related disease is associated with central pontine calcifications and atypical parkinsonism. Neurology: Genetics, 2020, 6, e399.	0.9	13
35	<scp>GGC</scp> Repeat Expansion in <scp>NOTCH2NLC</scp> Is Rare in European Leukoencephalopathy. Annals of Neurology, 2020, 88, 641-642.	2.8	14
36	GGC repeat expansion in NOTCH2NLC is rare in European patients with essential tremor. Brain, 2020, 143, e57-e57.	3.7	13

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

#	Article	IF	Citations
55	Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1230-1232.	0.9	18
56	Sequencing analysis of the SCA6 CAG expansion excludes an influence of repeat interruptions on disease onset. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1226-1227.	0.9	7
57	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
58	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. Neurobiology of Aging, 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. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E3601-E3603.	3.3	12
60	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
61	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. Frontiers in Cellular Neuroscience, 2018, 12, 429.	1.8	21
62	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
63	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1032.	4.9	15
64	LRP10 in Î±-synucleinopathies. Lancet Neurology, The, 2018, 17, 1033-1034.	4.9	11
65	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. Movement Disorders, 2018, 33, 1961-1965.	2.2	38
66	<scp>DNA</scp> repair in trinucleotide repeat ataxias. FEBS Journal, 2018, 285, 3669-3682.	2.2	12
67	Stratification of candidate genes for Parkinsonâ€™s disease using weighted protein-protein interaction network analysis. BMC Genomics, 2018, 19, 452.	1.2	35
68	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. JAMA Neurology, 2018, 75, 1416.	4.5	66
69	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
70	Mendelian randomization study shows no causal relationship between circulating urate levels and Parkinson's disease. Annals of Neurology, 2018, 84, 191-199.	2.8	43
71	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969.	1.1	34
72	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. Neurology, 2018, 90, e2059-e2067.	1.5	35

#	Article	IF	Citations
73	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. Cell, 2018, 173, 1705-1715.e16.	13.5	623
74	PREDICT-PD: An online approach to prospectively identify risk indicators of Parkinson's disease. Movement Disorders, 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. Parkinsonism and Related Disorders, 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. Neurobiology of Aging, 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. Parkinsonism and Related Disorders, 2017, 41, 37-43.	1.1	67
78	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. JAMA Neurology, 2017, 74, 780.	4.5	245
79	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. American Journal of Human Genetics, 2017, 100, 969-977.	2.6	38
80	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	9.4	186
81	Establishing the role of rare coding variants in known Parkinson's disease risk loci. Neurobiology of Aging, 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. Movement Disorders Clinical Practice, 2017, 4, 509-516.	0.8	35
83	Nonsyndromic Parkinson disease in a family with autosomal dominant optic atrophy due to OPA1 mutations. Neurology: Genetics, 2017, 3, e188.	0.9	27
84	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	3.7	323
85	Truncating mutations inSPASTpatients are associated with a high rate of psychiatric comorbidities in hereditary spastic paraplegia. Journal of Neurology, Neurosurgery and Psychiatry, 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. PLoS Medicine, 2017, 14, e1002314.	3.9	152
87	Variation in Recent Onset Parkinsonâ€™s Disease: Implications for Prodromal Detection. Journal of Parkinson's Disease, 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. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1183-1190.	0.9	24
89	B48â€…DNA repair pathways as a common genetic mechanism modulating the age at onset in polyglutamine diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A26.1-A26.	0.9	0
90	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

#	Article	IF	Citations
91	Triple trouble: a striking new phenotype or competing genes in a family with hereditary spastic paraplegia. Journal of Neurology, 2016, 263, 1232-1233.	1.8	4
92	Heterogeneity in clinical features and disease severity in ataxia-associated SYNE1 mutations. Journal of Neurology, 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. Parkinsonism and Related Disorders, 2016, 33, 96-101.	1.1	46
94	Late-onset Lafora disease with prominent parkinsonism due to a rare mutation in EPM2A. Neurology: Genetics, 2016, 2, e101.	0.9	16
95	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. Annals of Neurology, 2016, 79, 983-990.	2.8	183
96	A genome-wide association study in multiple system atrophy. Neurology, 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. Movement Disorders, 2016, 31, 1518-1526.	2.2	128
98	SLC25A46 mutations underlie progressive myoclonic ataxia with optic atrophy and neuropathy. Movement Disorders, 2016, 31, 1249-1251.	2.2	49
99	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
100	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. Brain, 2016, 139, 1904-1918.	3.7	170
101	Additional rare variant analysis in Parkinsonâ€™s disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. Human Molecular Genetics, 2016, 25, ddw348.	1.4	48
102	EFFECTS OF VASCULAR COMORBIDITY IN PARKINSON'S DISEASE. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, e1.13-e1.	0.9	0
103	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. American Journal of Human Genetics, 2016, 98, 1092-1100.	2.6	39
104	Olfaction inParkinsingle and compound heterozygotes in a cohort of young onset Parkinson's disease patients. Acta Neurologica Scandinavica, 2016, 134, 271-276.	1.0	21
105	Single-Molecule Imaging of Individual Amyloid Protein Aggregates in Human Biofluids. ACS Chemical Neuroscience, 2016, 7, 399-406.	1.7	99
106	Calcium is a key factor in Î±-synuclein induced neurotoxicity. Journal of Cell Science, 2016, 129, 1792-801.	1.2	136
107	Is the MC1R variant p.R160W associated with Parkinson's?. Annals of Neurology, 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. Lancet Neurology, The, 2016, 15, 585-596.	4.9	77

#	Article	IF	Citations
109	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
110	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
111	Polygenic risk of ischemic stroke is associated with cognitive ability. Neurology, 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. Antioxidants and Redox Signaling, 2016, 24, 376-391.	2.5	266
113	The genetic associations of acute anterior uveitis and their overlap with the genetics of ankylosing spondylitis. Genes and Immunity, 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. PLoS Medicine, 2016, 13, e1001976.	3.9	150
115	Ca2+ is a key factor in Î±-synuclein-induced neurotoxicity. Development (Cambridge), 2016, 143, e1.1-e1.1.	1.2	5
116	Tracking Parkinsonâ€™s: Study Design and Baseline Patient Data. Journal of Parkinson's Disease, 2015, 5, 947-959.	1.5	64
117	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	2.6	109
118	CHCHD2 and Parkinson's disease. Lancet Neurology, The, 2015, 14, 678-679.	4.9	50
119	Diagnostic clues and manifesting carriers in fukutin-related protein (FKRP) limb-girdle muscular dystrophy. Journal of the Neurological Sciences, 2015, 348, 266-268.	0.3	6
120	Genetic overlap between Alzheimerâ€™s disease and Parkinsonâ€™s disease at the MAPT locus. Molecular Psychiatry, 2015, 20, 1588-1595.	4.1	133
121	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. Neurology: Genetics, 2015, 1, e9.	0.9	90
122	Influence ofCOMTgenotype and affective distractors on the processing of self-generated thought. Social Cognitive and Affective Neuroscience, 2015, 10, 777-782.	1.5	11
123	Reply: Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2015, 138, e352-e352.	3.7	4
124	ADCY5 mutations are another cause of benign hereditary chorea. Neurology, 2015, 85, 80-88.	1.5	140
125	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. Neurobiology of Aging, 2015, 36, 1605.e7-1605.e12.	1.5	96
126	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

#	Article	IF	Citations
127	Mutations in HPCA Cause Autosomal-Recessive Primary Isolated Dystonia. American Journal of Human Genetics, 2015, 96, 657-665.	2.6	151
128	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
129	TheCACNA1BR1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. Human Molecular Genetics, 2015, 24, 5326-5329.	1.4	28
130	Preliminary investigation of the influence of dopamine regulating genes on social working memory. Social Neuroscience, 2014, 9, 437-451.	0.7	14
131	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2014, 95, 611-621.	2.6	89
132	PINK1 deficiency in Î²-cells increases basal insulin secretion and improves glucose tolerance in mice. Open Biology, 2014, 4, 140051.	1.5	40
133	ALS2 mutations. Neurology, 2014, 82, 1065-1067.	1.5	29
134	The phenotypic spectrum of DYT24 due to ANO3 mutations. Movement Disorders, 2014, 29, 928-934.	2.2	161
135	Hypersomnia with dilated pupils in adenosine monophosphate deaminase (<scp>AMPD</scp>) deficiency. Journal of Sleep Research, 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. Human Molecular Genetics, 2014, 23, 562-562.	1.4	5
137	Autosomal-recessive cerebellar ataxia caused by a novel ADCK3 mutation that elongates the protein: clinical, genetic and biochemical characterisation. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 493-498.	0.9	48
138	Susceptibility loci for pigmentation and melanoma in relation to Parkinson's disease. Neurobiology of Aging, 2014, 35, 1512.e5-1512.e10.	1.5	28
139	Advances in the Genetics of Parkinson's Disease: A Guide for the Clinician. Movement Disorders Clinical Practice, 2014, 1, 3-13.	0.8	17
140	Rare Individual Amyloid-Î² Oligomers Act on Astrocytes to Initiate Neuronal Damage. Biochemistry, 2014, 53, 2442-2453.	1.2	83
141	Analysis of Parkinson's disease brainâ€“derived DNA for alphaâ€synuclein coding somatic mutations. Movement Disorders, 2014, 29, 1060-1064.	2.2	22
142	Genetic comorbidities in Parkinson's disease. Human Molecular Genetics, 2014, 23, 831-841.	1.4	57
143	When the penny drops. Practical Neurology, 2014, 14, 409-414.	0.5	0
144	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. Neurology, 2014, 83, 1873-1875.	1.5	33

#	Article	IF	Citations
145	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
146	Parkinsonâ€™s disease in GTP cyclohydrolase 1 mutation carriers. Brain, 2014, 137, 2480-2492.	3.7	169
147	Screening of mutations in NOL3 in a myoclonic syndromes series. Journal of Neurology, 2014, 261, 1830-1831.	1.8	1
148	Assessment of Parkinson's disease risk loci in Greece. Neurobiology of Aging, 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. Nature Communications, 2014, 5, 4204.	5.8	72
150	Alpha-Synuclein Modulates [Ca2+]c of Neurons and Astrocytes that Trigger Cell Death. Biophysical Journal, 2014, 106, 529a.	0.2	0
151	Alpha-Synuclein Induces Mitochondrial Dysfunction Leading to a Higher Susceptibility of PTP Opening. Biophysical Journal, 2014, 106, 590a.	0.2	0
152	Friedreich's ataxia and other hereditary ataxias in Greece: An 18-year perspective. Journal of the Neurological Sciences, 2014, 336, 87-92.	0.3	13
153	A Genome-wide Association Analysis of a Broad Psychosis Phenotype Identifies Three Loci for Further Investigation. Biological Psychiatry, 2014, 75, 386-397.	0.7	44
154	No pathogenic GNAL mutations in 192 sporadic and familial cases of cervical dystonia. Movement Disorders, 2014, 29, 154-155.	2.2	8
155	The Parkinson's diseaseâ€“linked proteins Fbxo7 and Parkin interact to mediate mitophagy. Nature Neuroscience, 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. JAMA Neurology, 2013, 70, 1268-76.	4.5	51
157	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	9.4	1,395
158	The frequency of spinocerebellar ataxia type 23 in a UK population. Journal of Neurology, 2013, 260, 856-859.	1.8	12
159	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. Journal of Neurology, 2013, 260, 656-660.	1.8	17
160	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
161	A Novel Prion Disease Associated with Diarrhea and Autonomic Neuropathy. New England Journal of Medicine, 2013, 369, 1904-1914.	13.9	113
162	The Role of the Mitochondrial NCX in the Mechanism of Neurodegeneration in Parkinsonâ€™s Disease. Advances in Experimental Medicine and Biology, 2013, 961, 241-249.	0.8	25

#	Article	IF	Citations
163	Common variants in the HLA-DRB1â€“HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. Nature Genetics, 2013, 45, 208-213.	9.4	86
164	Mutational analysis of <scp>PMP22</scp>, <scp>EGR2</scp>, <scp>LITAF</scp> and <scp>NEFL</scp> in Greek Charcotâ€“Marieâ€“Tooth type 1 patients. Clinical Genetics, 2013, 83, 388-391.	1.0	2
165	Signalling properties of inorganic polyphosphate in the mammalian brain. Nature Communications, 2013, 4, 1362.	5.8	132
166	The genetics of dystonia: new twists in an old tale. Brain, 2013, 136, 2017-2037.	3.7	102
167	Ataxia telangiectasia presenting as dopa-responsive cervical dystonia. Neurology, 2013, 81, 1148-1151.	1.5	65
168	The Role of Interruptions in polyQ in the Pathology of SCA1. PLoS Genetics, 2013, 9, e1003648.	1.5	73
169	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 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. Human Molecular Genetics, 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. Human Molecular Genetics, 2013, 22, 1039-1049.	1.4	122
172	FAMILY HISTORY IN YOUNG ONSET PARKINSON'S DISEASE. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, e2.69-e2.	0.9	0
173	C9ORF72 expansions, parkinsonism, and Parkinson disease. Neurology, 2013, 81, 808-811.	1.5	57
174	Mutations in the autoregulatory domain of Î²â€tubulin 4a cause hereditary dystonia. Annals of Neurology, 2013, 73, 546-553.	2.8	148
175	TRACKING PARKINSON'S (THE PROBAND STUDY)â€“INTERIM REPORT FROM THE FIRST 1000 CASES. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, e2.70-e2.	0.9	0
176	Genotype and phenotype in Parkinson's disease: Lessons in heterogeneity from deep brain stimulation. Movement Disorders, 2013, 28, 1370-1375.	2.2	77
177	Fine-Mapping, Gene Expression and Splicing Analysis of the Disease Associated LRRK2 Locus. PLoS ONE, 2013, 8, e70724.	1.1	45
178	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. Human Molecular Genetics, 2012, 21, 4996-5009.	1.4	176
179	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	1.5	495
180	Myoclonus-dystonia syndrome due to tyrosine hydroxylase deficiency. Neurology, 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	PRRT2 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	Youngâ€onset parkinsonism due to homozygous duplication of Î±â€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 Î±-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 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
200	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
201	Mutations in ANO3 Cause Dominant Craniocervical Dystonia: Ion Channel Implicated in Pathogenesis. American Journal of Human Genetics, 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. Biological Psychiatry, 2012, 72, 620-628.	0.7	156
203	Analysis of spinocerebellar ataxias due to expanded triplet repeats in Greek patients with cerebellar ataxia. Journal of the Neurological Sciences, 2012, 318, 178-180.	0.3	7
204	Characterisation and Validation of Insertions and Deletions in 173 Patient Exomes. PLoS ONE, 2012, 7, e51292.	1.1	8
205	Genetic linkage analysis of a large family with photoparoxysmal response. Epilepsy Research, 2012, 99, 38-45.	0.8	1
206	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532.	2.2	108
207	Cooperative Genome-Wide Analysis Shows Increased Homozygosity in Early Onset Parkinson's Disease. PLoS ONE, 2012, 7, e28787.	1.1	21
208	Mutations in Nuclear Genes That Affect Mitochondrial Function in Parkinsonâ€™s Disease. , 2012, , 43-61.		0
209	Ataxia in a young patient. Practical Neurology, 2011, 11, 319-322.	0.5	0
210	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 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. Journal of the Neurological Sciences, 2011, 306, 62-65.	0.3	31
212	Mutational analysis of parkin and PINK1 in multiple system atrophy. Neurobiology of Aging, 2011, 32, 548.e5-548.e7.	1.5	16
213	Bioenergetic Consequences of PINK1 Mutations in Parkinson Disease. PLoS ONE, 2011, 6, e25622.	1.1	88
214	Interview: The genetics of Parkinsonâ€™s disease: piecing together the jigsaw. Neurodegenerative Disease Management, 2011, 1, 105-107.	1.2	0
215	LRRK2 expression in idiopathic and G2019S positive Parkinson's disease subjects: a morphological and quantitative study. Neuropathology and Applied Neurobiology, 2011, 37, 777-790.	1.8	44
216	Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. Nature Genetics, 2011, 43, 117-120.	9.4	390

#	Article	IF	Citations
217	Parkinson's disease and cancer: two wars, one front. Nature Reviews Cancer, 2011, 11, 813-823.	12.8	146
218	Mitophagy and Parkinson's disease: The PINK1â€“parkin link. Biochimica Et Biophysica Acta - Molecular Cell Research, 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. Lancet, The, 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. Nature Genetics, 2011, 43, 761-767.	9.4	778
221	An intragenic duplication in guanosine triphosphate cyclohydrolaseâ€1 gene in a dopaâ€responsive dystonia family. Movement Disorders, 2011, 26, 905-909.	2.2	12
222	Cell metabolism affects selective vulnerability in PINK1-associated Parkinson's disease. Journal of Cell Science, 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. Human Molecular Genetics, 2011, 20, 345-353.	1.4	202
224	PINK1 cleavage at position A103 by the mitochondrial protease PARL. Human Molecular Genetics, 2011, 20, 867-879.	1.4	385
225	A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease. PLoS Genetics, 2011, 7, e1002142.	1.5	247
226	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
227	Nonmotor symptoms in Parkin geneâ€related parkinsonism. Movement Disorders, 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. Movement Disorders, 2010, 25, 1506-1509.	2.2	21
229	Effects of age and MAOA genotype on the neural processing of social rejection. Genes, Brain and Behavior, 2010, 9, 628-637.	1.1	35
230	Genetic variability at the PARK16 locus. European Journal of Human Genetics, 2010, 18, 1356-1359.	1.4	85
231	Genetic and functional characterisation of the P/Q calcium channel in episodic ataxia with epilepsy. Journal of Physiology, 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. Nature Genetics, 2010, 42, 985-990.	9.4	918
233	Genome-wide association studies: the key to unlocking neurodegeneration?. Nature Neuroscience, 2010, 13, 789-794.	7.1	90
234	Endothelial, Sympathetic, and Cardiac Function in Inherited (6 R)- <scp>l</scp> -Erythro-5,6,7,8-Tetrahydro- <scp>l</scp> -Biopterin Deficiency. Circulation: Cardiovascular Genetics, 2010, 3, 513-522.	5.1	15

#	Article	IF	Citations
235	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. Brain, 2010, 133, 2136-2147.	3.7	132
236	Cancer and Neurodegeneration: Between the Devil and the Deep Blue Sea. PLoS Genetics, 2010, 6, e1001257.	1.5	141
237	Hyperexcitable Substantia Nigra Dopamine Neurons inPINK1- andHtrA2/Omi-Deficient Mice. Journal of Neurophysiology, 2010, 104, 3009-3020.	0.9	47
238	Targeting mitochondrial dysfunction in neurodegenerative disease: Part II. Expert Opinion on Therapeutic Targets, 2010, 14, 497-511.	1.5	58
239	Normal variation in fronto-occipital circuitry and cerebellar structure with an autism-associated polymorphism of CNTNAP2. NeuroImage, 2010, 53, 1030-1042.	2.1	105
240	Targeting mitochondrial dysfunction in neurodegenerative disease: Part I. Expert Opinion on Therapeutic Targets, 2010, 14, 369-385.	1.5	56
241	In vivo assessment of brain monoamine systems in parkin gene carriers: A PET study. Experimental Neurology, 2010, 222, 120-124.	2.0	25
242	Characterization of PLA2G6 as a locus for dystoniaâ€parkinsonism. Annals of Neurology, 2009, 65, 19-23.	2.8	399
243	Genetic Variants of the Î±-Synuclein Gene SNCA Are Associated with Multiple System Atrophy. PLoS ONE, 2009, 4, e7114.	1.1	144
244	GJB1 gene mutations in suspected inflammatory demyelinating neuropathies not responding to treatment. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 699-700.	0.9	29
245	A Genetically Mediated Bias in Decision Making Driven by Failure of Amygdala Control. Journal of Neuroscience, 2009, 29, 5985-5991.	1.7	183
246	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
247	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. Brain, 2009, 132, 1783-1794.	3.7	612
248	GLUCOCEREBROSIDASE MUTATIONS IN 108 NEUROPATHOLOGICALLY CONFIRMED CASES OF MULTIPLE SYSTEM ATROPHY. Neurology, 2009, 72, 1185-1186.	1.5	60
249	Differential DJ-1 gene expression in Parkinson's disease. Neurobiology of Disease, 2009, 36, 393-400.	2.1	42
250	SNCA variants are associated with increased risk for multiple system atrophy. Annals of Neurology, 2009, 65, 610-614.	2.8	257
251	PINK1 function in health and disease. EMBO Molecular Medicine, 2009, 1, 152-165.	3.3	125
252	Evidence for pre and postsynaptic nigrostriatal dysfunction in the fragile X tremorâ€“Ataxia syndrome. Movement Disorders, 2009, 24, 1245-1247.	2.2	15

#	Article	IF	Citations
253	Nigrostriatal dysfunction in homozygous and heterozygous parkin gene carriers: An ¹⁸Fâ€dopa PET progression study. Movement Disorders, 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. Cell Death and Differentiation, 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. Nature Cell Biology, 2009, 11, 1370-1375.	4.6	173
256	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
257	Genome-wide association study reveals genetic risk underlying Parkinson's disease. Nature Genetics, 2009, 41, 1308-1312.	9.4	1,745
258	No major role of common SV2A variation for predisposition or levetiracetam response in epilepsy. Epilepsy Research, 2009, 83, 44-51.	0.8	32
259	Association of MAPT haplotype-tagging SNPs with sporadic Parkinson's disease. Neurobiology of Aging, 2009, 30, 1477-1482.	1.5	48
260	PINK1-Associated Parkinson's Disease Is Caused by Neuronal Vulnerability to Calcium-Induced Cell Death. Molecular Cell, 2009, 33, 627-638.	4.5	584
261	Characterisation of a novel NR4A2 mutation in Parkinson's disease brain. Neuroscience Letters, 2009, 457, 75-79.	1.0	36
262	Cell Death Pathways in Parkinson's Disease: Role of Mitochondria. Antioxidants and Redox Signaling, 2009, 11, 2135-2149.	2.5	70
263	Riluzole treatment, survival and diagnostic criteria in Parkinson plus disorders: The NNIPPS Study. Brain, 2009, 132, 156-171.	3.7	298
264	Molecular basis of Parkinson's disease. NeuroReport, 2009, 20, 150-156.	0.6	69
265	Clinical and genetic analysis of spinocerebellar ataxia type 11. Cerebellum, 2008, 7, 159-164.	1.4	23
266	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
267	Motor cortical physiology in patients and asymptomatic carriers of parkin gene mutations. Movement Disorders, 2008, 23, 1812-1819.	2.2	29
268	Mitochondrial function and morphology are impaired in parkinâ€mutant fibroblasts. Annals of Neurology, 2008, 64, 555-565.	2.8	339
269	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
270	What Have PINK1 and HtrA2 Genes Told Us about the Role of Mitochondria in Parkinson's Disease?. Annals of the New York Academy of Sciences, 2008, 1147, 30-36.	1.8	22

#	Article	IF	Citations
271	Clinical heterogeneity and genotypeâ€“phenotype correlations in hereditary spastic paraplegia because of Spatacsin mutations (SPG11). European Journal of Neurology, 2008, 15, 1065-1070.	1.7	34
272	Genetic testing in neurology. Medicine, 2008, 36, 566-568.	0.2	0
273	PINK1 Is Necessary for Long Term Survival and Mitochondrial Function in Human Dopaminergic Neurons. PLoS ONE, 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. Neuropathology and Applied Neurobiology, 2008, 34, 492-505.	1.8	28
275	Second consensus statement on the diagnosis of multiple system atrophy. Neurology, 2008, 71, 670-676.	1.5	2,720
276	Genetics of progressive supranuclear palsy. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2008, 89, 475-485.	1.0	0
277	Neuropathology of primary adult-onset dystonia. Neurology, 2008, 70, 695-699.	1.5	42
278	Mutations in the HSP27 (HSPB1) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. Neurology, 2008, 71, 1660-1668.	1.5	168
279	Hyposmia in G2019S LRRK2-related parkinsonism. Neurology, 2008, 71, 1021-1026.	1.5	82
280	Multiple mitochondrial DNA deletions in monozygotic twins with OPMD. Journal of Neurology, Neurosurgery and Psychiatry, 2008, 79, 68-71.	0.9	13
281	Clinical and genetic analysis of spinocerebellar ataxia type 11. Cerebellum, 2008, 7, 1-6.	1.4	0
282	Mitochondrial ND5 Gene Variation Associated with Encephalomyopathy and Mitochondrial ATP Consumption. Journal of Biological Chemistry, 2007, 282, 36845-36852.	1.6	59
283	Deletion at ITPR1 Underlies Ataxia in Mice and Spinocerebellar Ataxia 15 in Humans. PLoS Genetics, 2007, 3, e108.	1.5	269
284	Large-scale pathways-based association study in amyotrophic lateral sclerosis. Brain, 2007, 130, 2292-2301.	3.7	32
285	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
286	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
287	Alternative ion channel splicing in mesial temporal lobe epilepsy and Alzheimer's disease. Genome Biology, 2007, 8, R32.	13.9	38
288	The mitochondrial protease HtrA2 is regulated by Parkinson's disease-associated kinase PINK1. Nature Cell Biology, 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. Nature Genetics, 2007, 39, 1434-1436.	9.4	185
290	A Multicenter Study of BRD2 as a Risk Factor for Juvenile Myoclonic Epilepsy. Epilepsia, 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. Lancet Neurology, The, 2007, 6, 970-980.	4.9	175
292	Understanding the molecular causes of Parkinson's disease. Trends in Molecular Medicine, 2006, 12, 521-528.	3.5	274
293	Altered cleavage and localization of PINK1 to aggresomes in the presence of proteasomal stress. Journal of Neurochemistry, 2006, 98, 156-169.	2.1	146
294	Expanding insights of mitochondrial dysfunction in Parkinson's disease. Nature Reviews Neuroscience, 2006, 7, 207-219.	4.9	773
295	A pharmacogenetic exploration of vigabatrin-induced visual field constriction. Epilepsy Research, 2006, 70, 144-152.	0.8	26
296	Examining the role of common genetic variation in the Î³2 subunit of the GABAA receptor in epilepsy using tagging SNPs. Epilepsy Research, 2006, 70, 229-238.	0.8	17
297	Nigral degeneration and striatal dopaminergic dysfunction in idiopathic andparkin-linked Parkinson's disease. Movement Disorders, 2006, 21, 299-305.	2.2	18
298	NR4A2 genetic variation in sporadic Parkinson's disease: A genewide approach. Movement Disorders, 2006, 21, 1960-1963.	2.2	15
299	TheADH1C stop mutation in multiple system atrophy patients and healthy probands in the United Kingdom and Germany. Movement Disorders, 2006, 21, 2034-2034.	2.2	6
300	UCHL-1is not a Parkinson's disease susceptibility gene. Annals of Neurology, 2006, 59, 627-633.	2.8	123
301	A heterozygous effect for PINK1 mutations in Parkinson's disease?. Annals of Neurology, 2006, 60, 414-419.	2.8	149
302	Familial dopa-responsive cervical dystonia. Neurology, 2006, 66, 599-601.	1.5	38
303	PINK1 protein in normal human brain and Parkinson's disease. Brain, 2006, 129, 1720-1731.	3.7	291
304	Genetic association studies of complex neurological diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 1302-1304.	0.9	19
305	Mitochondria in Parkinson Disease. Archives of Neurology, 2006, 63, 649.	4.9	27
306	The Â-synuclein gene in multiple system atrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 77, 464-467.	0.9	45

#	Article	IF	Citations
307	The syndrome of (predominantly cervical) dystonia and cerebellar ataxia: new cases indicate a distinct but heterogeneous entity. Journal of Neurology, Neurosurgery and Psychiatry, 2006, 78, 774-775.	0.9	19
308	Molecular genetic pathways in Parkinson's disease: a review. Clinical Science, 2005, 109, 355-364.	1.8	37
309	A single-nucleotide polymorphism tagging set for human drug metabolism and transport. Nature Genetics, 2005, 37, 84-89.	9.4	142
310	PARK11 is not linked with Parkinson's disease in European families. European Journal of Human Genetics, 2005, 13, 193-197.	1.4	23
311	Apolipoprotein E4 is probably responsible for the chromosome 19 linkage peak for Parkinson's disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 136B, 72-74.	1.1	27
312	Parkin disease in a Brazilian kindred: Manifesting heterozygotes and clinical follow-up over 10 years. Movement Disorders, 2005, 20, 479-484.	2.2	32
313	Parkinsonism and nigrostriatal dysfunction are associated with spinocerebellar ataxia type 6 (SCA6). Movement Disorders, 2005, 20, 1115-1119.	2.2	45
314	UCHL-1 gene in multiple system atrophy: A haplotype tagging approach. Movement Disorders, 2005, 20, 1338-1343.	2.2	17
315	Commentary on â€œA genome wide linkage disequilibrium screen in Parkinsonâ€™s diseaseâ€• Journal of Neurology, 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. Journal of Medical Genetics, 2005, 42, 837-846.	1.5	225
317	Population genetic approaches to neurological disease: Parkinson's disease as an example. Philosophical Transactions of the Royal Society B: Biological Sciences, 2005, 360, 1573-1578.	1.8	4
318	Introduction: genetic variation and human health. Philosophical Transactions of the Royal Society B: Biological Sciences, 2005, 360, 1539-1541.	1.8	2
319	Failure to replicate previously reported genetic associations with sporadic temporal lobe epilepsy: where to from here?. Brain, 2005, 128, 1832-1840.	3.7	87
320	Association of genetic loci: Replication or not, that is the question. Neurology, 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. Proceedings of the National Academy of Sciences of the United States of America, 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. Journal of Medical Genetics, 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. Brain, 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. Brain, 2005, 128, 2786-2796.	3.7	315

#	Article	IF	Citations
325	A common LRRK2 mutation in idiopathic Parkinson's disease. Lancet, The, 2005, 365, 415-416.	6.3	391
326	Molecular pathogenesis of Parkinson's disease. Human Molecular Genetics, 2005, 14, 2749-2755.	1.4	187
327	Synphilin-1 and parkin show overlapping expression patterns in human brain and form aggresomes in response to proteasomal inhibition. Neurobiology of Disease, 2005, 20, 401-411.	2.1	40
328	A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. Brain, 2004, 127, 973-980.	3.7	77
329	The structure of the tau haplotype in controls and in progressive supranuclear palsy. Human Molecular Genetics, 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. Journal of Medical Genetics, 2004, 41, 900-907.	1.5	38
331	Autosomal dominant cerebellar ataxia: SCA2 is the most frequent mutation in eastern India. Journal of Neurology, Neurosurgery and Psychiatry, 2004, 75, 448-452.	0.9	62
332	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
333	Genome-Wide Analysis of the Parkinsonism-Dementia Complex of Guam. Archives of Neurology, 2004, 61, 1889-97.	4.9	44
334	The expression of DJ-1 (PARK7) in normal human CNS and idiopathic Parkinson's disease. Brain, 2004, 127, 420-430.	3.7	404
335	Trinucleotide repeats and neurodegenerative disease. Brain, 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. Brain, 2004, 127, 2657-2671.	3.7	493
337	Identifying Candidate Causal Variants Responsible for Altered Activity of the ABCB1 Multidrug Resistance Gene. Genome Research, 2004, 14, 1333-1344.	2.4	107
338	Striatal and cortical pre- and postsynaptic dopaminergic dysfunction in sporadic parkin-linked parkinsonism. Brain, 2004, 127, 1332-1342.	3.7	104
339	Assessment of a DJ-1 (PARK7) polymorphism in Finnish PD. Neurology, 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. European Journal of Neuroscience, 2004, 20, 2038-2048.	1.2	17
341	PINK, PANK, or PARK? A clinicians' guide to familial parkinsonism. Lancet Neurology, The, 2004, 3, 652-662.	4.9	61
342	Genetic causes of Parkinson?s disease: UCHL-1. Cell and Tissue Research, 2004, 318, 189-194.	1.5	51

#	Article	IF	Citations
343	Causes of Parkinson?s disease: genetics of DJ-1. Cell and Tissue Research, 2004, 318, 185-188.	1.5	51
344	Genetic Approaches to Solving Common Diseases. Journal of Neurology, 2004, 251, 1169-1172.	1.8	1
345	A functional polymorphism regulating dopamine ?-hydroxylase influences against Parkinson's disease. Annals of Neurology, 2004, 55, 443-446.	2.8	59
346	The gene responsible forPARK6 Parkinson's disease,PINK1, does not influence common forms of parkinsonism. Annals of Neurology, 2004, 56, 329-335.	2.8	38
347	Ataxin-7 aggregation and ubiquitination in infantile SCA7 with 180 CAG repeats. Annals of Neurology, 2004, 56, 448-452.	2.8	47
348	Connexin 32 promoter P2 mutations: A mechanism of peripheral nerve dysfunction. Annals of Neurology, 2004, 56, 730-734.	2.8	51
349	Population genetics for target identification. Drug Discovery Today: Technologies, 2004, 1, 69-74.	4.0	10
350	Hereditary Early-Onset Parkinson's Disease Caused by Mutations in PINK1. Science, 2004, 304, 1158-1160.	6.0	3,060
351	Cloning of the Gene Containing Mutations that Cause PARK8-Linked Parkinson's Disease. Neuron, 2004, 44, 595-600.	3.8	2,183
352	Somatic and germline mosaicism in sporadic early-onset Alzheimer's disease. Human Molecular Genetics, 2004, 13, 1219-1224.	1.4	93
353	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
354	Slowly progressive cerebellar ataxia and cervical dystonia: Clinical presentation of a new form of spinocerebellar ataxia?. Movement Disorders, 2003, 18, 200-206.	2.2	35
355	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
356	How much phenotypic variation can be attributed toparkingenotype?. Annals of Neurology, 2003, 54, 176-185.	2.8	271
357	The role of pathogenicDJ-1 mutations in Parkinson's disease. Annals of Neurology, 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. Neuropathology and Applied Neurobiology, 2003, 29, 288-302.	1.8	194
359	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
360	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

#	Article	IF	Citations
361	Different patterns of electrophysiological deficits in manifesting and non-manifesting carriers of the DYT1 gene mutation. Brain, 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. Human Molecular Genetics, 2003, 13, 117-135.	1.4	72
363	Gluten ataxia in perspective: epidemiology, genetic susceptibility and clinical characteristics. Brain, 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. Human Molecular Genetics, 2003, 12, 1917-1925.	1.4	51
365	Parkin disease: a phenotypic study of a large case series. Brain, 2003, 126, 1279-1292.	3.7	427
366	Parkin mutations are frequent in patients with isolated earlyâ€onset parkinsonism. Brain, 2003, 126, 1271-1278.	3.7	279
367	Autosomal recessive, DYT2-like primary torsion dystonia. Neurology, 2003, 61, 1801-1803.	1.5	122
368	Six novel connexin32 (GJB1) mutations in X-linked Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 304-306.	0.9	56
369	Sequence analysis of tau in familial and sporadic progressive supranuclear palsy. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 72, 388-390.	0.9	17
370	Running a neurogenetic clinic. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 2ii-4.	0.9	8
371	Genetics of movement disorders and ataxia. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 73, 22ii-26.	0.9	37
372	Neuronal intranuclear inclusions in SCA2: a genetic, morphological and immunohistochemical study of two cases. Brain, 2002, 125, 656-663.	3.7	87
373	Intrafamilial Phenotypic Variability in Friedreich Ataxia Associated With a G130V Mutation in the FRDA Gene. Archives of Neurology, 2002, 59, 296.	4.9	17
374	Clinical and genetic characterization of families with triple A (Allgrove) syndrome. Brain, 2002, 125, 2681-2690.	3.7	137
375	Familial adult onset of Krabbe's disease resembling hereditary spastic paraplegia with normal neuroimaging. Journal of Neurology, Neurosurgery and Psychiatry, 2002, 72, 635-638.	0.9	43
376	Dopa-Responsive Dystonia - The Story so Far. Neuropediatrics, 2002, 33, 1-5.	0.3	60
377	Immunological study of hereditary motor and sensory neuropathy type 1a (HMSN1a). Journal of Neurology, Neurosurgery and Psychiatry, 2002, 72, 230-235.	0.9	37
378	The tau locus is not significantly associated with pathologically confirmed sporadic Parkinson's disease. Neuroscience Letters, 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. Brain, 2002, 125, 2248-2256.	3.7	141
380	Mouse models for neurological disease. Lancet Neurology, The, 2002, 1, 215-224.	4.9	41
381	Complex relationship between Parkin mutations and Parkinson disease. American Journal of Medical Genetics Part A, 2002, 114, 584-591.	2.4	193
382	Park6-linked parkinsonism occurs in several european families. Annals of Neurology, 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. Annals of Neurology, 2002, 51, 740-749.	2.8	40
384	Myoclonus-dystonia syndrome: Îµ-sarcoglycan mutations and phenotype. Annals of Neurology, 2002, 52, 489-492.	2.8	143
385	Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: An18F-dopa PET study. Annals of Neurology, 2002, 52, 849-853.	2.8	192
386	Corticobasal degeneration syndrome with basal ganglia calcification: Fahr's disease as a corticobasal look-alike?. Movement Disorders, 2002, 17, 563-567.	2.2	27
387	Genetic and clinical heterogeneity in paroxysmal kinesigenic dyskinesia: Evidence for a third EKD gene. Movement Disorders, 2002, 17, 717-725.	2.2	85
388	PARK6 is a common cause of familial parkinsonism. Neurological Sciences, 2002, 23, s117-s118.	0.9	34
389	Mutation analysis of the sodium/hydrogen exchanger gene (NHE5) in familial paroxysmal kinesigenic dyskinesia. Journal of Neural Transmission, 2002, 109, 1189-1194.	1.4	12
390	Mutation of the sterol 27-hydroxylase gene (CYP27A1) in a Taiwanese family with cerebrotendinous xanthomatosis. Journal of Neurology, 2002, 249, 1311-1312.	1.8	8
391	Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. European Journal of Human Genetics, 2002, 10, 773-781.	1.4	172
392	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
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. American Journal of Human Genetics, 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. American Journal of Human Genetics, 2001, 68, 895-900.	2.6	459
395	Localization of the Gene for Distal Hereditary Motor Neuronopathy VII (dHMN-VII) to Chromosome 2q14. American Journal of Human Genetics, 2001, 68, 1270-1276.	2.6	68
396	No pathogenic mutations in the synphilin-1 gene in Parkinson's disease. Neuroscience Letters, 2001, 307, 125-127.	1.0	18

#	Article	IF	Citations
397	Strong association of a novel Tau promoter haplotype in progressive supranuclear palsy. Neuroscience Letters, 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. Neuroscience Letters, 2001, 312, 118-120.	1.0	26
399	Chromosome 13 dementia syndromes as models of neurodegeneration. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 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. Neurological Sciences, 2001, 22, 95-96.	0.9	23
401	Parkinson's disease is not associated with the combined ?-synuclein/apolipoprotein E susceptibility genotype. Annals of Neurology, 2001, 49, 665-668.	2.8	66
402	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
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. Annals of Neurology, 2001, 49, 362-366.	2.8	118
404	A conserved sorting-associated protein is mutant in chorea-acanthocytosis. Nature Genetics, 2001, 28, 119-120.	9.4	357
405	Systemic Amyloid Deposits in Familial British Dementia. Journal of Biological Chemistry, 2001, 276, 43909-43914.	1.6	73
406	Cervical dystonia is associated with a polymorphism in the dopamine (D5) receptor gene. Journal of Neurology, Neurosurgery and Psychiatry, 2001, 71, 262-264.	0.9	50
407	DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.13â€“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-? concentrations. Annals of Neurology, 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. Movement Disorders, 2000, 15, 30-35.	2.2	37
412	Paroxysmal dystonic choreoathetosis: Clinical features and investigation of pathophysiology in a large family. Movement Disorders, 2000, 15, 648-657.	2.2	46
413	Large, expanded repeats in SCA8 are not confined to patients with cerebellar ataxia. Nature Genetics, 2000, 24, 214-215.	9.4	109
414	A new family with paroxysmal exercise induced dystonia and migraine: a clinical and genetic study. Journal of Neurology, Neurosurgery and Psychiatry, 2000, 68, 609-614.	0.9	72

#	Article	IF	Citations
415	Mitochondrial DNA Point Mutation T9176C in Leigh Syndrome. Journal of Child Neurology, 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. Parkinsonism and Related Disorders, 2000, 6, 195-197.	1.1	61
417	The genetics of Parkinson's disease. Current Opinion in Genetics and Development, 2000, 10, 292-298.	1.5	73
418	Association between Early-Onset Parkinson's Disease and Mutations in theParkinGene. New England Journal of Medicine, 2000, 342, 1560-1567.	13.9	1,448
419	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PSâ€1 mutations that lead to exceptionally high amyloidâ€î² concentrations. Annals of Neurology, 2000, 48, 806-808.	2.8	3
420	Clinical genetics of familial progressive supranuclear palsy. Brain, 1999, 122, 1233-1245.	3.7	124
421	Huntington's disease progression. Brain, 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. Brain, 1999, 122, 817-825.	3.7	314
423	Phenotypic variation of a new P0 mutation in genetically identical twins. Journal of Neurology, 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. Annals of Neurology, 1999, 46, 916-919.	2.8	129
425	Neurofibrillary tangle parkinsonian disorders?tau pathology andtau genetics. Movement Disorders, 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. American Journal of Human Genetics, 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. American Journal of Human Genetics, 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. American Journal of Human Genetics, 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. Neuroscience Letters, 1999, 270, 1-4.	1.0	75
430	Mutations in the gene encoding human persyn are not associated with amyotrophic lateral sclerosis or familial Parkinson's disease. Neuroscience Letters, 1999, 274, 21-24.	1.0	16
431	Prader-Willi and Angelman syndromes: update on genetic mechanisms and diagnostic complexities. Current Opinion in Neurology, 1999, 12, 149-154.	1.8	23
432	Low frequency of pathogenic mutations in the ubiquitin carboxyterminal hydrolase gene in familial ParkinsonÊ¼s disease. NeuroReport, 1999, 10, 427-429.	0.6	119

#	Article	IF	Citations
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 ParkinsonÊ¼s disease. Current Opinion in Neurology, 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. Annals of Neurology, 1998, 43, 680-683.	2.8	42
436	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
437	Dopa-responsive dystonia: A clinical and molecular genetic study. Annals of Neurology, 1998, 44, 649-656.	2.8	153
438	Genetic aspects of Parkinson's disease. Movement Disorders, 1998, 13, 203-211.	2.2	38
439	Generalized chorea in two patients harboring the Friedreich's ataxia gene trinucleotide repeat expansion. Movement Disorders, 1998, 13, 339-340.	2.2	48
440	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
441	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
442	De Novo Expansion of Intermediate Alleles in Spinocerebellar Ataxia 7. Human Molecular Genetics, 1998, 7, 1809-1813.	1.4	96
443	Fine mapping of de novo CMT1A and HNPP rearrangements within CMTIA-REPs evidences two distinct sex-dependent mechanisms and candidate sequences involved in recombination. Human Molecular Genetics, 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). Human Molecular Genetics, 1998, 7, 751-753.	1.4	98
445	Genetic risk factors in parkinson's disease. Annals of Neurology, 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. Brain, 1998, 121, 459-467.	3.7	84
447	Paroxysmal dystonic choreoathetosis. Genetic linkage studies in a British family. Brain, 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. Brain, 1997, 120, 673-680.	3.7	77
449	Association of slow acetylator genotype for N-acetyltransferase 2 with familial Parkinson's disease. Lancet, The, 1997, 350, 1136-1139.	6.3	121
450	Chorea-Acanthocytosis: Genetic Linkage to Chromosome 9q21. American Journal of Human Genetics, 1997, 61, 899-908.	2.6	126

#	Article	IF	Citations
451	Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion. Nature Genetics, 1997, 17, 65-70.	9.4	758
452	Sexâ€“dependent rearrangements resulting in CMT1A and HNPP. Nature Genetics, 1997, 17, 136-137.	9.4	57
453	Depletion of mitochondrial DNA by ddC in untransformed human cell lines. Somatic Cell and Molecular Genetics, 1997, 23, 287-290.	0.7	45
454	Mitochondrial DNA polymorphisms in pathologically proven Parkinsonâ€™s disease. Journal of Neurology, 1997, 244, 262-265.	1.8	50
455	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
456	Progressive cognitive decline with truncal/limb ataxia and ballistic movements. Movement Disorders, 1997, 12, 1075-1084.	2.2	0
457	The GTP-cyclohydrolase I gene in atypical Parkinsonian patients: a clinico-genetic study. Journal of the Neurological Sciences, 1996, 141, 27-32.	0.3	23
458	The human homologue of the weaver mouse gene in familial and sporadic Parkinson's disease. Neuroscience, 1996, 72, 877-879.	1.1	23
459	Mitochondrial disorders in neuro-ophthalmology. Current Opinion in Neurology, 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. Human Molecular Genetics, 1996, 5, 403-406.	1.4	107
461	Genes and susceptibility to multiple sclerosis. Acta Neurologica Scandinavica, 1995, 91, 43-51.	1.0	124
462	Susceptibility to multiple sclerosis and the immunoglobulin heavy chain variable region. Journal of Neurology, 1995, 242, 677-682.	1.8	36
463	Multiple sclerosis and the HLA-D region: linkage and association studies. Journal of Neuroimmunology, 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.. Journal of Neurology, Neurosurgery and Psychiatry, 1994, 57, 1191-1194.	0.9	33
465	No linkage between multiple sclerosis and the T cell receptor Î± chain locus. Journal of the Neurological Sciences, 1994, 124, 32-37.	0.3	13
466	The pathogenesis of demyelinating disease. Progress in Neurobiology, 1994, 43, 143-173.	2.8	53
467	Multiple sclerosis in the Cambridge health district of east Anglia.. Journal of Neurology, Neurosurgery and Psychiatry, 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 â€“ what it really means and where next. , 0, , 1-5.		1
472	Channelopathies. , 0, , 121-135.		0