Nicholas W. Wood

List of Publications by Year in Descending Order

472 papers	53,188 citations	111 h-index	221 g-index
499 ext. papers	61,517 ext. citations	9.8 avg, IF	6.78 L-index

#	Paper	IF	Citations
472	Cerebellar and Midbrain Lysosomal Enzyme Deficiency in Isolated Dystonia.. *Movement Disorders*, **2022**,	7	1
471	The role of body fat in multiple sclerosis susceptibility and severity: A Mendelian randomisation study.. *Multiple Sclerosis Journal*, **2022**, 13524585221092644	5	0
470	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	36.3	19
469	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
468	Dissecting the Phenotype and Genotype of PLA2G6-Related Parkinsonism. *Movement Disorders*, **2021**,	7	8
467	Mitochondrial DNA Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. *Annals of Neurology*, **2021**, 89, 1240-1247	9.4	3
466	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. *Journal of Clinical Investigation*, **2021**, 131,	15.9	2
465	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. *JAMA Neurology*, **2021**, 78, 464-472	17.2	17
464	Mendelian Randomisation Finds No Causal Association between Urate and Parkinson's Disease Progression. *Movement Disorders*, **2021**, 36, 2182-2187	7	1
463	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. *Annals of Neurology*, **2021**, 90, 35-42	9.4	6
462	Childhood-Onset Chorea Caused by a Recurrent De Novo DRD2 Variant. *Movement Disorders*, **2021**, 36, 1472-1473	7	2
461	Identification of sixteen novel candidate genes for late onset Parkinson's disease. *Molecular Neurodegeneration*, **2021**, 16, 35	19	3
460	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. *Movement Disorders*, **2021**, 36, 251-255	7	11
459	Genome-Wide Association Studies of Cognitive and Motor Progression in Parkinson's Disease. *Movement Disorders*, **2021**, 36, 424-433	7	27
458	Analysis of DNM3 and VAMP4 as genetic modifiers of LRRK2 Parkinson's disease. *Neurobiology of Aging*, **2021**, 97, 148.e17-148.e24	5.6	9
457	NOTCH2NLC Intermediate-Length Repeat Expansion and Parkinson's Disease in Patients of European Descent. *Annals of Neurology*, **2021**, 89, 633-635	9.4	3
456	Expanding the Spectrum of AP5Z1-Related Hereditary Spastic Paraplegia (HSP-SPG48): A Multicenter Study on a Rare Disease. *Movement Disorders*, **2021**, 36, 1034-1038	7	4

455	-related ataxia is a mimic of early multiple system atrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**,	5.5	11
454	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. *Annals of Neurology*, **2021**, 90, 193-202	9.4	11
453	Spastic paraplegia preceding -related familial Alzheimer's disease. *Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring*, **2021**, 13, e12186	5.2	1
452	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. *Nature Communications*, **2021**, 12, 7342	17.4	2
451	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,	6.3	11
450	-related disease is associated with central pontine calcifications and atypical parkinsonism. *Neurology: Genetics*, **2020**, 6, e399	3.8	7
449	GGC Repeat Expansion in NOTCH2NLC Is Rare in European Leukoencephalopathy. *Annals of Neurology*, **2020**, 88, 641-642	9.4	5
448	GGC repeat expansion in NOTCH2NLC is rare in European patients with essential tremor. *Brain*, **2020**, 143, e57	11.2	9
447	The repeat variant in MSH3 is not a genetic modifier for spinocerebellar ataxia type 3 and Friedreich's ataxia. *Brain*, **2020**, 143, e25	11.2	2
446	The influence of microsatellite polymorphisms in sex steroid receptor genes ESR1, ESR2 and AR on sex differences in brain structure. *NeuroImage*, **2020**, 221, 117087	7.9	2
445	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. *Movement Disorders*, **2020**, 35, 774-780	7	27
444	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	11.5	29
443	RFC1 Intronic Repeat Expansions Absent in Pathologically Confirmed Multiple Systems Atrophy. *Movement Disorders*, **2020**, 35, 1277-1279	7	13
442	LRRK2 activation controls the repair of damaged endomembranes in macrophages. *EMBO Journal*, **2020**, 39, e104494	13	37
441	Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. *Brain*, **2020**, 143, 234-248	11.2	69
440	Neuronal intranuclear inclusion disease is genetically heterogeneous. *Annals of Clinical and Translational Neurology*, **2020**, 7, 1716-1725	5.3	18
439	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	8.1	16
438	Dopa-Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansions. *Movement Disorders*, **2020**, 35, 1890-1891	7	0

437	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. *Brain*, **2020**, 143, 2771-2787	11.2	20
436	Using Mendelian randomization to understand and develop treatments for neurodegenerative disease. *Brain Communications*, **2020**, 2, fcaa031	4.5	4
435	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	7.5	0
434	Loss-of-Function Variants in HOPS Complex Genes VPS16 and VPS41 Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. *Annals of Neurology*, **2020**, 88, 867-877	9.4	33
433	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. *European Journal of Human Genetics*, **2020**, 28, 1763-1768	5.3	5
432	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. *Movement Disorders*, **2019**, 34, 1851-1863	7	18
431	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. *Movement Disorders*, **2019**, 34, 460-468	7	40
430	PDXK mutations cause polyneuropathy responsive to pyridoxal 5'-phosphate supplementation. *Annals of Neurology*, **2019**, 86, 225-240	9.4	18
429	L-dopa responsiveness in early Parkinson's disease is associated with the rate of motor progression. *Parkinsonism and Related Disorders*, **2019**, 65, 55-61	3.6	10
428	Delineating the phenotype of autosomal-recessive HPCA mutations: Not only isolated dystonia!. *Movement Disorders*, **2019**, 34, 589-592	7	4
427	Population-based identity-by-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	3.5	2
426	Genetic analysis of Mendelian mutations in a large UK population-based Parkinson's disease study. *Brain*, **2019**, 142, 2828-2844	11.2	35
425	The Parkinson's Disease Mendelian Randomization Research Portal. *Movement Disorders*, **2019**, 34, 1864-1872	7	29
424	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	24.1	562
423	loss of function causes autosomal recessive spastic ataxia and optic atrophy. *Annals of Clinical and Translational Neurology*, **2018**, 5, 216-221	5.3	8
422	Genotype-phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. *Movement Disorders*, **2018**, 33, 1119-1129	7	17
421	Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1230-1232	5.5	14
420	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	5.5	4

419	Features of -associated Parkinson's disease at presentation in the UK study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 702-709	5.5	55
418	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. *Neurobiology of Aging*, **2018**, 64, 159.e5-159.e8	5.6	23
417	Genetic variation in 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	11.5	9
416	Genome, transcriptome and proteome: the rise of omics data and their integration in biomedical sciences. *Briefings in Bioinformatics*, **2018**, 19, 286-302	13.4	293
415	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. *JAMA Neurology*, **2018**, 75, 1416-1422	17.2	50
414	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
413	Mendelian randomization study shows no causal relationship between circulating urate levels and Parkinson's disease. *Annals of Neurology*, **2018**, 84, 191-199	9.4	29
412	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. *Human Mutation*, **2018**, 39, 965-969	4.7	27
411	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. *Neurology*, **2018**, 90, e2059-e2067	6.5	25
410	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. *Cell*, **2018**, 173, 1705-1715.e16	56.2	360
409	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. *Frontiers in Cellular Neuroscience*, **2018**, 12, 429	6.1	11
408	Developing and validating Parkinson's disease subtypes and their motor and cognitive progression. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 1279-1287	5.5	66
407	LRP10 in synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1032	24.1	14
406	LRP10 in synucleinopathies. *Lancet Neurology, The*, **2018**, 17, 1033-1034	24.1	9
405	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. *Movement Disorders*, **2018**, 33, 1961-1965	7	18
404	DNA repair in trinucleotide repeat ataxias. *FEBS Journal*, **2018**, 285, 3669-3682	5.7	7
403	Stratification of candidate genes for Parkinson's disease using weighted protein-protein interaction network analysis. *BMC Genomics*, **2018**, 19, 452	4.5	17
402	PREDICT-PD: An online approach to prospectively identify risk indicators of Parkinson's disease. *Movement Disorders*, **2017**, 32, 219-226	7	42

401	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	3.6	9
400	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	5.6	54
399	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. *Parkinsonism and Related Disorders*, **2017**, 41, 37-43	3.6	54
398	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. *JAMA Neurology*, **2017**, 74, 780-792	17.2	150
397	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. *American Journal of Human Genetics*, **2017**, 100, 969-977	11	25
396	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. *Nature Genetics*, **2017**, 49, 223-237	36.3	116
395	Establishing the role of rare coding variants in known Parkinson's disease risk loci. *Neurobiology of Aging*, **2017**, 59, 220.e11-220.e18	5.6	11
394	Autonomic Dysfunction in Early Parkinson's Disease: Results from the United Kingdom Tracking Parkinson's Study. *Movement Disorders Clinical Practice*, **2017**, 4, 509-516	2.2	18
393	Nonsyndromic Parkinson disease in a family with autosomal dominant optic atrophy due to mutations. *Neurology: Genetics*, **2017**, 3, e188	3.8	20
392	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. *Brain*, **2017**, 140, 3191-3203	11.2	209
391	Truncating mutations in patients are associated with a high rate of psychiatric comorbidities in hereditary spastic paraplegia. *Journal of Neurology, Neurosurgery and Psychiatry*, **2017**, 88, 681-687	5.5	18
390	Estimating the causal influence of body mass index on risk of Parkinson disease: A Mendelian randomisation study. *PLoS Medicine*, **2017**, 14, e1002314	11.6	93
389	Cerebellar Ataxias and Related Conditions **2016**, 685-698		
388	A genome-wide association study in multiple system atrophy. *Neurology*, **2016**, 87, 1591-1598	6.5	104
387	Vascular disease and vascular risk factors in relation to motor features and cognition in early Parkinson's disease. *Movement Disorders*, **2016**, 31, 1518-1526	7	90
386	SLC25A46 mutations underlie progressive myoclonic ataxia with optic atrophy and neuropathy. *Movement Disorders*, **2016**, 31, 1249-51	7	37
385	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1043-8	36.3	328
384	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. *Brain*, **2016**, 139, 1904-18	11.2	123

383	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. *Human Molecular Genetics*, **2016**, 25, 5483-5489	5.6	40
382	EFFECTS OF VASCULAR COMORBIDITY IN PARKINSON'S DISEASE. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, e1.13-e1	5.5	
381	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. *American Journal of Human Genetics*, **2016**, 98, 1092-1100	11	30
380	Olfaction in Parkin single and compound heterozygotes in a cohort of young onset Parkinson's disease patients. *Acta Neurologica Scandinavica*, **2016**, 134, 271-6	3.8	20
379	Single-Molecule Imaging of Individual Amyloid Protein Aggregates in Human Biofluids. *ACS Chemical Neuroscience*, **2016**, 7, 399-406	5.7	75
378	Ca2+ is a key factor in □synuclein-induced neurotoxicity. *Journal of Cell Science*, **2016**, 129, 1792-801	5.3	106
377	Is the MC1R variant p.R160W associated with Parkinson's?. *Annals of Neurology*, **2016**, 79, 159-61	9.4	14
376	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. *Lancet Neurology, The*, **2016**, 15, 585-96	24.1	59
375	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	11.5	130
374	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	11	225
373	Polygenic risk of ischemic stroke is associated with cognitive ability. *Neurology*, **2016**, 86, 611-8	6.5	13
372	Alpha-Synuclein Oligomers Interact with Metal Ions to Induce Oxidative Stress and Neuronal Death in Parkinson's Disease. *Antioxidants and Redox Signaling*, **2016**, 24, 376-91	8.4	192
371	The genetic associations of acute anterior uveitis and their overlap with the genetics of ankylosing spondylitis. *Genes and Immunity*, **2016**, 17, 46-51	4.4	27
370	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	11.6	100
369	Ca2+ is a key factor in □synuclein-induced neurotoxicity. *Development (Cambridge)*, **2016**, 143, e1.1-e1.1	6.6	3
368	Variation in Recent Onset Parkinson's Disease: Implications for Prodromal Detection. *Journal of Parkinson's Disease*, **2016**, 6, 289-300	5.3	18
367	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	5.5	19
366	B48 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	5.5	

365	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. *American Journal of Human Genetics*, **2016**, 98, 763-71	11	74
364	Triple trouble: a striking new phenotype or competing genes in a family with hereditary spastic paraplegia. *Journal of Neurology*, **2016**, 263, 1232-3	5.5	2
363	Heterogeneity in clinical features and disease severity in ataxia-associated SYNE1 mutations. *Journal of Neurology*, **2016**, 263, 1503-10	5.5	21
362	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	3.6	36
361	Late-onset Lafora disease with prominent parkinsonism due to a rare mutation in EPM2A. *Neurology: Genetics*, **2016**, 2, e101	3.8	11
360	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. *Annals of Neurology*, **2016**, 79, 983-90	9.4	135
359	Genetic overlap between Alzheimer's disease and Parkinson's disease at the MAPT locus. *Molecular Psychiatry*, **2015**, 20, 1588-95	15.1	107
358	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. *Neurology: Genetics*, **2015**, 1, e9	3.8	63
357	Influence of COMT genotype and affective distractors on the processing of self-generated thought. *Social Cognitive and Affective Neuroscience*, **2015**, 10, 777-82	4	10
356	Reply: Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. *Brain*, **2015**, 138, e352	11.2	4
355	ADCY5 mutations are another cause of benign hereditary chorea. *Neurology*, **2015**, 85, 80-8	6.5	63
354	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. *Neurobiology of Aging*, **2015**, 36, 1605.e7-12	5.6	70
353	Polygenic risk of Parkinson disease is correlated with disease age at onset. *Annals of Neurology*, **2015**, 77, 582-91	9.4	77
352	Mutations in HPCA cause autosomal-recessive primary isolated dystonia. *American Journal of Human Genetics*, **2015**, 96, 657-65	11	59
351	Structural characterization of toxic oligomers that are kinetically trapped during α-synuclein fibril formation. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, E1994-2003	11.5	278
350	The CACNA1B R1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. *Human Molecular Genetics*, **2015**, 24, 5326-9	5.6	19
349	Tracking Parkinson's: Study Design and Baseline Patient Data. *Journal of Parkinson's Disease*, **2015**, 5, 947-59	5.3	31
348	A missense mutation in KCTD17 causes autosomal dominant myoclonus-dystonia. *American Journal of Human Genetics*, **2015**, 96, 938-47	11	77

347	CHCHD2 and Parkinson's disease. *Lancet Neurology, The*, **2015**, 14, 678-9	24.1	43
346	Diagnostic clues and manifesting carriers in fukutin-related protein (FKRP) limb-girdle muscular dystrophy. *Journal of the Neurological Sciences*, **2015**, 348, 266-8	3.2	5
345	Advances in the Genetics of Parkinson's Disease: A Guide for the Clinician. *Movement Disorders Clinical Practice*, **2014**, 1, 3-13	2.2	10
344	Rare individual amyloid-β oligomers act on astrocytes to initiate neuronal damage. *Biochemistry*, **2014**, 53, 2442-53	3.2	68
343	Analysis of Parkinson's disease brain-derived DNA for alpha-synuclein coding somatic mutations. *Movement Disorders*, **2014**, 29, 1060-4	7	18
342	Genetic comorbidities in Parkinson's disease. *Human Molecular Genetics*, **2014**, 23, 831-41	5.6	49
341	When the penny drops. *Practical Neurology*, **2014**, 14, 409-14	2.4	
340	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. *Neurology*, **2014**, 83, 1873-5	6.5	29
339	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. *Nature Genetics*, **2014**, 46, 989-93	36.3	1261
338	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. *Brain*, **2014**, 137, 2480-92	11.2	127
337	Screening of mutations in NOL3 in a myoclonic syndromes series. *Journal of Neurology*, **2014**, 261, 1830-1	5.5	1
336	Assessment of Parkinson's disease risk loci in Greece. *Neurobiology of Aging*, **2014**, 35, 442.e9-442.e16	5.6	12
335	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. *Nature Communications*, **2014**, 5, 4204	17.4	54
334	Friedreich's ataxia and other hereditary ataxias in Greece: an 18-year perspective. *Journal of the Neurological Sciences*, **2014**, 336, 87-92	3.2	7
333	A genome-wide association analysis of a broad psychosis phenotype identifies three loci for further investigation. *Biological Psychiatry*, **2014**, 75, 386-97	7.9	36
332	No pathogenic GNAL mutations in 192 sporadic and familial cases of cervical dystonia. *Movement Disorders*, **2014**, 29, 154-5	7	7
331	Preliminary investigation of the influence of dopamine regulating genes on social working memory. *Social Neuroscience*, **2014**, 9, 437-51	2	11
330	Mutations in SNX14 cause a distinctive autosomal-recessive cerebellar ataxia and intellectual disability syndrome. *American Journal of Human Genetics*, **2014**, 95, 611-21	11	73

329	PINK1 deficiency in β-cells increases basal insulin secretion and improves glucose tolerance in mice. *Open Biology*, **2014**, 4, 140051	7	32
328	ALS2 mutations: juvenile amyotrophic lateral sclerosis and generalized dystonia. *Neurology*, **2014**, 82, 1065-7	6.5	19
327	The phenotypic spectrum of DYT24 due to ANO3 mutations. *Movement Disorders*, **2014**, 29, 928-34	7	67
326	Hypersomnia with dilated pupils in adenosine monophosphate deaminase (AMPD) deficiency. *Journal of Sleep Research*, **2014**, 23, 118-20	5.8	1
325	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. *Human Molecular Genetics*, **2014**, 23, 562-562	5.6	3
324	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, 493-8	5.5	38
323	Susceptibility loci for pigmentation and melanoma in relation to Parkinson's disease. *Neurobiology of Aging*, **2014**, 35, 1512.e5-1512.e10	5.6	25
322	The Parkinson's disease-linked proteins Fbxo7 and Parkin interact to mediate mitophagy. *Nature Neuroscience*, **2013**, 16, 1257-65	25.5	220
321	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	17.2	46
320	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. *Nature Genetics*, **2013**, 45, 1150-9	36.3	1153
319	The frequency of spinocerebellar ataxia type 23 in a UK population. *Journal of Neurology*, **2013**, 260, 856-9	5.5	12
318	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. *Journal of Neurology*, **2013**, 260, 656-60	5.5	15
317	The glucocerobrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. *Movement Disorders*, **2013**, 28, 232-236	7	86
316	A novel prion disease associated with diarrhea and autonomic neuropathy. *New England Journal of Medicine*, **2013**, 369, 1904-14	59.2	93
315	The role of the mitochondrial NCX in the mechanism of neurodegeneration in Parkinson's disease. *Advances in Experimental Medicine and Biology*, **2013**, 961, 241-9	3.6	20
314	Common variants in the HLA-DRB1-HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. *Nature Genetics*, **2013**, 45, 208-13	36.3	76
313	Mutational analysis of PMP22, EGR2, LITAF and NEFL in Greek Charcot-Marie-Tooth type 1 patients. *Clinical Genetics*, **2013**, 83, 388-91	4	2
312	Signalling properties of inorganic polyphosphate in the mammalian brain. *Nature Communications*, **2013**, 4, 1362	17.4	103

311	The genetics of dystonia: new twists in an old tale. *Brain*, **2013**, 136, 2017-37	11.2	80
310	Ataxia telangiectasia presenting as dopa-responsive cervical dystonia. *Neurology*, **2013**, 81, 1148-51	6.5	52
309	The role of interruptions in polyQ in the pathology of SCA1. *PLoS Genetics*, **2013**, 9, e1003648	6	46
308	Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus. *Human Molecular Genetics*, **2013**, 22, 4653-60	5.6	24
307	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. *Human Molecular Genetics*, **2013**, 22, 1039-49	5.6	96
306	FAMILY HISTORY IN YOUNG ONSET PARKINSON'S DISEASE. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, e2.69-e2	5.5	
305	C9ORF72 expansions, parkinsonism, and Parkinson disease: a clinicopathologic study. *Neurology*, **2013**, 81, 808-11	6.5	49
304	Mutations in the autoregulatory domain of β-tubulin 4a cause hereditary dystonia. *Annals of Neurology*, **2013**, 73, 546-53	9.4	114
303	TRACKING PARKINSON'S (THE PROBAND STUDY): INTERIM REPORT FROM THE FIRST 1000 CASES. *Journal of Neurology, Neurosurgery and Psychiatry*, **2013**, 84, e2.70-e2	5.5	
302	Genotype and phenotype in Parkinson's disease: lessons in heterogeneity from deep brain stimulation. *Movement Disorders*, **2013**, 28, 1370-5	7	55
301	Fine-mapping, gene expression and splicing analysis of the disease associated LRRK2 locus. *PLoS ONE*, **2013**, 8, e70724	3.7	40
300	Genetic linkage analysis of a large family with photoparoxysmal response. *Epilepsy Research*, **2012**, 99, 38-45	3	
299	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. *Movement Disorders*, **2012**, 27, 526-32	7	80
298	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. *Neurology*, **2012**, 79, 127-31	6.5	29
297	Use of support vector machines for disease risk prediction in genome-wide association studies: concerns and opportunities. *Human Mutation*, **2012**, 33, 1708-18	4.7	28
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