

# Nicholas W. Wood

## List of Publications by Citations

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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	Hereditary early-onset Parkinson's disease caused by mutations in PINK1. <i>Science</i> , <b>2004</b> , 304, 1158-60	33.3	2586
471	Second consensus statement on the diagnosis of multiple system atrophy. <i>Neurology</i> , <b>2008</b> , 71, 670-6	6.5	2069
470	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. <i>Nature</i> , <b>2011</b> , 476, 214-9	50.4	1948
469	Cloning of the gene containing mutations that cause PARK8-linked Parkinson's disease. <i>Neuron</i> , <b>2004</b> , 44, 595-600	13.9	1860
468	Genome-wide association study reveals genetic risk underlying Parkinson's disease. <i>Nature Genetics</i> , <b>2009</b> , 41, 1308-12	36.3	1469
467	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. <i>Nature Genetics</i> , <b>2014</b> , 46, 989-93	36.3	1261
466	Association between early-onset Parkinson's disease and mutations in the parkin gene. <i>New England Journal of Medicine</i> , <b>2000</b> , 342, 1560-7	59.2	1242
465	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. <i>Nature Genetics</i> , <b>2013</b> , 45, 1150-9	36.3	1153
464	Phenotype, genotype, and worldwide genetic penetrance of LRRK2-associated Parkinson's disease: a case-control study. <i>Lancet Neurology</i> , <b>2008</b> , 7, 583-90	24.1	1075
463	A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1. <i>Nature Genetics</i> , <b>2010</b> , 42, 985-90	36.3	773
462	Imputation of sequence variants for identification of genetic risks for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet</i> , <b>2011</b> , 377, 641-9	40	733
461	Cloning of the SCA7 gene reveals a highly unstable CAG repeat expansion. <i>Nature Genetics</i> , <b>1997</b> , 17, 65-70	36.3	694
460	Expanding insights of mitochondrial dysfunction in Parkinson's disease. <i>Nature Reviews Neuroscience</i> , <b>2006</b> , 7, 207-19	13.5	686
459	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. <i>Nature Genetics</i> , <b>2012</b> , 44, 1341-8	36.3	681
458	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , <b>2018</b> , 360,	33.3	666
457	Interaction between ERAP1 and HLA-B27 in ankylosing spondylitis implicates peptide handling in the mechanism for HLA-B27 in disease susceptibility. <i>Nature Genetics</i> , <b>2011</b> , 43, 761-7	36.3	646
456	Association of multidrug resistance in epilepsy with a polymorphism in the drug-transporter gene ABCB1. <i>New England Journal of Medicine</i> , <b>2003</b> , 348, 1442-8	59.2	611

455	Direct observation of the interconversion of normal and toxic forms of $\alpha$ -synuclein. <i>Cell</i> , <b>2012</b> , 149, 1048-56.2	58.2	588
454	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet Neurology</i> , <b>2019</b> , 18, 1091-1102	24.1	562
453	PINK1-associated Parkinson's disease is caused by neuronal vulnerability to calcium-induced cell death. <i>Molecular Cell</i> , <b>2009</b> , 33, 627-38	17.6	507
452	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. <i>Brain</i> , <b>2009</b> , 132, 1783-94	11.2	488
451	Comprehensive research synopsis and systematic meta-analyses in Parkinson's disease genetics: The PDGene database. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002548	6	420
450	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. <i>Nature Genetics</i> , <b>2009</b> , 41, 1330-4	36.3	411
449	Localization of a novel locus for autosomal recessive early-onset parkinsonism, PARK6, on human chromosome 1p35-p36. <i>American Journal of Human Genetics</i> , <b>2001</b> , 68, 895-900	11	389
448	The mitochondrial protease HtrA2 is regulated by Parkinson's disease-associated kinase PINK1. <i>Nature Cell Biology</i> , <b>2007</b> , 9, 1243-52	23.4	386
447	Glucocerebrosidase deficiency in substantia nigra of parkinson disease brains. <i>Annals of Neurology</i> , <b>2012</b> , 72, 455-63	9.4	384
446	Genomic Dissection of Bipolar Disorder and Schizophrenia, Including 28 Subphenotypes. <i>Cell</i> , <b>2018</b> , 173, 1705-1715.e16	56.2	360
445	The spectrum of pathological involvement of the striatonigral and olivopontocerebellar systems in multiple system atrophy: clinicopathological correlations. <i>Brain</i> , <b>2004</b> , 127, 2657-71	11.2	359
444	A robust model for read count data in exome sequencing experiments and implications for copy number variant calling. <i>Bioinformatics</i> , <b>2012</b> , 28, 2747-54	7.2	345
443	The expression of DJ-1 (PARK7) in normal human CNS and idiopathic Parkinson's disease. <i>Brain</i> , <b>2004</b> , 127, 420-30	11.2	341
442	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , <b>2016</b> , 48, 1043-8	36.3	328
441	Characterization of PLA2G6 as a locus for dystonia-parkinsonism. <i>Annals of Neurology</i> , <b>2009</b> , 65, 19-23	9.4	320
440	The role of pathogenic DJ-1 mutations in Parkinson's disease. <i>Annals of Neurology</i> , <b>2003</b> , 54, 283-6	9.4	320
439	Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. <i>Nature Genetics</i> , <b>2011</b> , 43, 117-20	36.3	319
438	Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke. <i>Nature Genetics</i> , <b>2012</b> , 44, 328-33	36.3	314

437	PINK1 cleavage at position A103 by the mitochondrial protease PARL. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 867-79	5.6	314
436	Parkin disease: a phenotypic study of a large case series. <i>Brain</i> , <b>2003</b> , 126, 1279-92	11.2	312
435	Genome, transcriptome and proteome: the rise of omics data and their integration in biomedical sciences. <i>Briefings in Bioinformatics</i> , <b>2018</b> , 19, 286-302	13.4	293
434	Mutations in the gene LRRK2 encoding dardarin (PARK8) cause familial Parkinson's disease: clinical, pathological, olfactory and functional imaging and genetic data. <i>Brain</i> , <b>2005</b> , 128, 2786-96	11.2	283
433	A common LRRK2 mutation in idiopathic Parkinson's disease. <i>Lancet, The</i> , <b>2005</b> , 365, 415-6	4.0	283
432	A conserved sorting-associated protein is mutant in chorea-acanthocytosis. <i>Nature Genetics</i> , <b>2001</b> , 28, 119-20	36.3	282
431	Mitochondrial function and morphology are impaired in parkin-mutant fibroblasts. <i>Annals of Neurology</i> , <b>2008</b> , 64, 555-65	9.4	280
430	Structural characterization of toxic oligomers that are kinetically trapped during $\beta$ -synuclein fibril formation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2015</b> , 112, E1994-2003	11.5	278
429	Genetic predictors of the maximum doses patients receive during clinical use of the anti-epileptic drugs carbamazepine and phenytoin. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2005</b> , 102, 5507-12	11.5	278
428	PINK1 protein in normal human brain and Parkinson's disease. <i>Brain</i> , <b>2006</b> , 129, 1720-31	11.2	267
427	Targeting amyloid-beta in glaucoma treatment. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2007</b> , 104, 13444-9	11.5	262
426	A novel mutation in the human voltage-gated potassium channel gene (Kv1.1) associates with episodic ataxia type 1 and sometimes with partial epilepsy. <i>Brain</i> , <b>1999</b> , 122 ( Pt 5), 817-25	11.2	257
425	PINK1 is necessary for long term survival and mitochondrial function in human dopaminergic neurons. <i>PLoS ONE</i> , <b>2008</b> , 3, e2455	3.7	252
424	Riluzole treatment, survival and diagnostic criteria in Parkinson plus disorders: the NNIPPS study. <i>Brain</i> , <b>2009</b> , 132, 156-71	11.2	248
423	Parkin mutations are frequent in patients with isolated early-onset parkinsonism. <i>Brain</i> , <b>2003</b> , 126, 1271-8	11.2	245
422	Understanding the molecular causes of Parkinson's disease. <i>Trends in Molecular Medicine</i> , <b>2006</b> , 12, 521-8	11.5	240
421	SNCA variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , <b>2009</b> , 65, 610-4	9.4	232
420	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 500-513	11.1	225

4 <sup>19</sup>	How much phenotypic variation can be attributed to parkin genotype?. <i>Annals of Neurology</i> , <b>2003</b> , 54, 176-85	9.4	224
4 <sup>18</sup>	Deletion at ITPR1 underlies ataxia in mice and spinocerebellar ataxia 15 in humans. <i>PLoS Genetics</i> , <b>2007</b> , 3, e108	6	221
4 <sup>17</sup>	The Parkinson's disease-linked proteins Fbxo7 and Parkin interact to mediate mitophagy. <i>Nature Neuroscience</i> , <b>2013</b> , 16, 1257-65	25.5	220
4 <sup>16</sup>	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , <b>2017</b> , 140, 3191-3203	11.2	209
4 <sup>15</sup>	A two-stage meta-analysis identifies several new loci for Parkinson's disease. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1002142	6	209
4 <sup>14</sup>	Rare deletions at 16p13.11 predispose to a diverse spectrum of sporadic epilepsy syndromes. <i>American Journal of Human Genetics</i> , <b>2010</b> , 86, 707-18	11	206
4 <sup>13</sup>	Mutations in the gene PRRT2 cause paroxysmal kinesigenic dyskinesia with infantile convulsions. <i>Cell Reports</i> , <b>2012</b> , 1, 2-12	10.6	205
4 <sup>12</sup>	Alpha-Synuclein Oligomers Interact with Metal Ions to Induce Oxidative Stress and Neuronal Death in Parkinson's Disease. <i>Antioxidants and Redox Signaling</i> , <b>2016</b> , 24, 376-91	8.4	192
4 <sup>11</sup>	Linkage disequilibrium fine mapping and haplotype association analysis of the tau gene in progressive supranuclear palsy and corticobasal degeneration. <i>Journal of Medical Genetics</i> , <b>2005</b> , 42, 837-46	5.8	189
4 <sup>10</sup>	Huntington's disease progression. PET and clinical observations. <i>Brain</i> , <b>1999</b> , 122 ( Pt 12), 2353-63	11.2	181
4 <sup>09</sup>	Clinical and subclinical dopaminergic dysfunction in PARK6-linked parkinsonism: an 18F-dopa PET study. <i>Annals of Neurology</i> , <b>2002</b> , 52, 849-53	9.4	179
4 <sup>08</sup>	Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 345-53	5.6	178
4 <sup>07</sup>	Gluten ataxia in perspective: epidemiology, genetic susceptibility and clinical characteristics. <i>Brain</i> , <b>2003</b> , 126, 685-91	11.2	175
4 <sup>06</sup>	Mutations in ANO3 cause dominant craniocervical dystonia: ion channel implicated in pathogenesis. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 1041-50	11	172
4 <sup>05</sup>	Homozygous deletions in parkin gene in European and North African families with autosomal recessive juvenile parkinsonism. The European Consortium on Genetic Susceptibility in Parkinson's Disease and the French Parkinson's Disease Genetics Study Group. <i>Lancet, The</i> , <b>1998</b> , 352, 1355-6	40	171
4 <sup>04</sup>	Selection and evaluation of tagging SNPs in the neuronal-sodium-channel gene SCN1A: implications for linkage-disequilibrium gene mapping. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 551-65	11	171
4 <sup>03</sup>	Pathological inclusion bodies in tauopathies contain distinct complements of tau with three or four microtubule-binding repeat domains as demonstrated by new specific monoclonal antibodies. <i>Neuropathology and Applied Neurobiology</i> , <b>2003</b> , 29, 288-302	5.2	167
4 <sup>02</sup>	A genetically mediated bias in decision making driven by failure of amygdala control. <i>Journal of Neuroscience</i> , <b>2009</b> , 29, 5985-91	6.6	165

401	Molecular pathogenesis of Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 2749-55	5.6	162
400	Complex relationship between Parkin mutations and Parkinson disease. <i>American Journal of Medical Genetics Part A</i> , <b>2002</b> , 114, 584-91		154
399	Mutations in TTBK2, encoding a kinase implicated in tau phosphorylation, segregate with spinocerebellar ataxia type 11. <i>Nature Genetics</i> , <b>2007</b> , 39, 1434-6	36.3	152
398	Multicentre search for genetic susceptibility loci in sporadic epilepsy syndrome and seizure types: a case-control study. <i>Lancet Neurology, The</i> , <b>2007</b> , 6, 970-80	24.1	152
397	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. <i>JAMA Neurology</i> , <b>2017</b> , 74, 780-792	17.2	150
396	Transcriptional repression of p53 by parkin and impairment by mutations associated with autosomal recessive juvenile Parkinson's disease. <i>Nature Cell Biology</i> , <b>2009</b> , 11, 1370-5	23.4	147
395	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 4996-5009	5.6	145
394	Mitophagy and Parkinson's disease: the PINK1-parkin link. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>2011</b> , 1813, 623-33	4.9	144
393	Dopa-responsive dystonia: a clinical and molecular genetic study. <i>Annals of Neurology</i> , <b>1998</b> , 44, 649-56	9.4	143
392	Mutations in the HSP27 (HSPB1) gene cause dominant, recessive, and sporadic distal HMN/CMT type 2. <i>Neurology</i> , <b>2008</b> , 71, 1660-8	6.5	142
391	Mutational spectrum of the CHAC gene in patients with chorea-acanthocytosis. <i>European Journal of Human Genetics</i> , <b>2002</b> , 10, 773-81	5.3	140
390	Common variants at the MHC locus and at chromosome 16q24.1 predispose to Barrett's esophagus. <i>Nature Genetics</i> , <b>2012</b> , 44, 1131-6	36.3	139
389	Mitochondrial dysfunction triggered by loss of HtrA2 results in the activation of a brain-specific transcriptional stress response. <i>Cell Death and Differentiation</i> , <b>2009</b> , 16, 449-64	12.7	137
388	Altered cleavage and localization of PINK1 to aggresomes in the presence of proteasomal stress. <i>Journal of Neurochemistry</i> , <b>2006</b> , 98, 156-69	6	136
387	Genome scans and candidate gene approaches in the study of common diseases and variable drug responses. <i>Trends in Genetics</i> , <b>2003</b> , 19, 615-22	8.5	136
386	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. <i>Annals of Neurology</i> , <b>2016</b> , 79, 983-90	9.4	135
385	A single-nucleotide polymorphism tagging set for human drug metabolism and transport. <i>Nature Genetics</i> , <b>2005</b> , 37, 84-9	36.3	134
384	PRRT2 gene mutations: from paroxysmal dyskinesia to episodic ataxia and hemiplegic migraine. <i>Neurology</i> , <b>2012</b> , 79, 2115-21	6.5	132

383	Kinetic model of the aggregation of alpha-synuclein provides insights into prion-like spreading. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2016</b> , 113, E1206-15	11.5	130
382	Genome-wide association study implicates HLA-C*01:02 as a risk factor at the major histocompatibility complex locus in schizophrenia. <i>Biological Psychiatry</i> , <b>2012</b> , 72, 620-8	7.9	130
381	Genetic variants of the alpha-synuclein gene SNCA are associated with multiple system atrophy. <i>PLoS ONE</i> , <b>2009</b> , 4, e7114	3.7	130
380	A heterozygous effect for PINK1 mutations in Parkinson's disease?. <i>Annals of Neurology</i> , <b>2006</b> , 60, 414-9	9.4	130
379	Trinucleotide repeats and neurodegenerative disease. <i>Brain</i> , <b>2004</b> , 127, 2385-405	11.2	128
378	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , <b>2014</b> , 137, 2480-92	11.2	127
377	Autosomal dominant cerebellar ataxia type III: linkage in a large British family to a 7.6-cM region on chromosome 15q14-21.3. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 420-6	11	127
376	Autosomal-dominant GTPCH1-deficient DRD: clinical characteristics and long-term outcome of 34 patients. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2009</b> , 80, 839-45	5.5	125
375	Cytochrome c oxidase deficiency associated with the first stop-codon point mutation in human mtDNA. <i>American Journal of Human Genetics</i> , <b>1998</b> , 63, 29-36	11	124
374	Genetic and phenotypic characterization of complex hereditary spastic paraplegia. <i>Brain</i> , <b>2016</b> , 139, 1904-18	11.8	123
373	Cortical alpha-synuclein load is associated with amyloid-beta plaque burden in a subset of Parkinson's disease patients. <i>Acta Neuropathologica</i> , <b>2008</b> , 115, 417-25	14.3	121
372	Progression of nigrostriatal dysfunction in a parkin kindred: an [18F]dopa PET and clinical study. <i>Brain</i> , <b>2002</b> , 125, 2248-56	11.2	120
371	Parkinson's disease and cancer: two wars, one front. <i>Nature Reviews Cancer</i> , <b>2011</b> , 11, 812-23	31.3	119
370	The mitochondrial DNA G13513A transition in ND5 is associated with a LHON/MELAS overlap syndrome and may be a frequent cause of MELAS. <i>Annals of Neurology</i> , <b>1999</b> , 46, 916-9	9.4	119
369	Different patterns of electrophysiological deficits in manifesting and non-manifesting carriers of the DYT1 gene mutation. <i>Brain</i> , <b>2003</b> , 126, 2074-80	11.2	118
368	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , <b>2017</b> , 49, 223-237	36.3	116
367	Common genetic variation and susceptibility to partial epilepsies: a genome-wide association study. <i>Brain</i> , <b>2010</b> , 133, 2136-47	11.2	115
366	Regional distribution of amyloid-Bri deposition and its association with neurofibrillary degeneration in familial British dementia. <i>American Journal of Pathology</i> , <b>2001</b> , 158, 515-26	5.8	115



365	Mutations in the autoregulatory domain of $\beta$ -tubulin 4a cause hereditary dystonia. <i>Annals of Neurology</i> , <b>2013</b> , 73, 546-53	9.4	114
364	Myoclonus-dystonia syndrome: epsilon-sarcoglycan mutations and phenotype. <i>Annals of Neurology</i> , <b>2002</b> , 52, 489-92	9.4	114
363	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid- $\beta$ concentrations. <i>Annals of Neurology</i> , <b>2000</b> , 48, 806-808	8.4	113
362	Chorea-acanthocytosis: genetic linkage to chromosome 9q21. <i>American Journal of Human Genetics</i> , <b>1997</b> , 61, 899-908	11	111
361	Clinical and genetic characterization of families with triple A (Allgrove) syndrome. <i>Brain</i> , <b>2002</b> , 125, 2681-90	11.2	111
360	Genes and susceptibility to multiple sclerosis. <i>Acta Neurologica Scandinavica</i> , <b>1995</b> , 161, 43-51	3.8	111
359	Association of slow acetylator genotype for N-acetyltransferase 2 with familial Parkinson's disease. <i>Lancet, The</i> , <b>1997</b> , 350, 1136-9	4.0	110
358	Systematic review and UK-based study of PARK2 (parkin), PINK1, PARK7 (DJ-1) and LRRK2 in early-onset Parkinson's disease. <i>Movement Disorders</i> , <b>2012</b> , 27, 1522-9	7	109
357	Genetic overlap between Alzheimer's disease and Parkinson's disease at the MAPT locus. <i>Molecular Psychiatry</i> , <b>2015</b> , 20, 1588-95	15.1	107
356	UCHL-1 is not a Parkinson's disease susceptibility gene. <i>Annals of Neurology</i> , <b>2006</b> , 59, 627-33	9.4	107
355	Clinical genetics of familial progressive supranuclear palsy. <i>Brain</i> , <b>1999</b> , 122 ( Pt 7), 1233-45	11.2	107
354	Ca <sup>2+</sup> is a key factor in $\beta$ -synuclein-induced neurotoxicity. <i>Journal of Cell Science</i> , <b>2016</b> , 129, 1792-801	5.3	106
353	Cancer and neurodegeneration: between the devil and the deep blue sea. <i>PLoS Genetics</i> , <b>2010</b> , 6, e1001257	15.7	106
352	A genome-wide association study in multiple system atrophy. <i>Neurology</i> , <b>2016</b> , 87, 1591-1598	6.5	104
351	An mtDNA mutation in the initiation codon of the cytochrome C oxidase subunit II gene results in lower levels of the protein and a mitochondrial encephalomyopathy. <i>American Journal of Human Genetics</i> , <b>1999</b> , 64, 1330-9	11	104
350	Signalling properties of inorganic polyphosphate in the mammalian brain. <i>Nature Communications</i> , <b>2013</b> , 4, 1362	17.4	103
349	The structure of the tau haplotype in controls and in progressive supranuclear palsy. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1267-74	5.6	102
348	Low frequency of pathogenic mutations in the ubiquitin carboxy-terminal hydrolase gene in familial Parkinson's disease. <i>NeuroReport</i> , <b>1999</b> , 10, 427-9	1.7	102



347	Tau gene and Parkinson's disease: a case-control study and meta-analysis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2004</b> , 75, 962-5	5.5	100
346	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. <i>PLoS Medicine</i> , <b>2016</b> , 13, e1001976	11.6	100
345	Large, expanded repeats in SCA8 are not confined to patients with cerebellar ataxia. <i>Nature Genetics</i> , <b>2000</b> , 24, 214-5	36.3	99
344	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 1039-49	5.6	96
343	A novel TRK A (NTRK1) mutation associated with hereditary sensory and autonomic neuropathy type V. <i>Annals of Neurology</i> , <b>2001</b> , 49, 521-525	9.4	96
342	PINK1 function in health and disease. <i>EMBO Molecular Medicine</i> , <b>2009</b> , 1, 152-65	12	95
341	Identifying candidate causal variants responsible for altered activity of the ABCB1 multidrug resistance gene. <i>Genome Research</i> , <b>2004</b> , 14, 1333-44	9.7	94
340	A novel prion disease associated with diarrhea and autonomic neuropathy. <i>New England Journal of Medicine</i> , <b>2013</b> , 369, 1904-14	59.2	93
339	Origin of the mutations in the parkin gene in Europe: exon rearrangements are independent recurrent events, whereas point mutations may result from Founder effects. <i>American Journal of Human Genetics</i> , <b>2001</b> , 68, 617-26	11	93
338	Estimating the causal influence of body mass index on risk of Parkinson disease: A Mendelian randomisation study. <i>PLoS Medicine</i> , <b>2017</b> , 14, e1002314	11.6	93
337	Striatal and cortical pre- and postsynaptic dopaminergic dysfunction in sporadic parkin-linked parkinsonism. <i>Brain</i> , <b>2004</b> , 127, 1332-42	11.2	92
336	The fragile X tremor ataxia syndrome in the differential diagnosis of multiple system atrophy: data from the EMSA Study Group. <i>Brain</i> , <b>2005</b> , 128, 1855-60	11.2	91
335	Vascular disease and vascular risk factors in relation to motor features and cognition in early Parkinson's disease. <i>Movement Disorders</i> , <b>2016</b> , 31, 1518-1526	7	90
334	Nova2 interacts with a cis-acting polymorphism to influence the proportions of drug-responsive splice variants of SCN1A. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 876-83	11	90
333	DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.13B6.32 in an Italian family with cranial-cervical or upper limb onset. <i>Annals of Neurology</i> , <b>2001</b> , 49, 362-366	9.4	90
332	Normal variation in fronto-occipital circuitry and cerebellar structure with an autism-associated polymorphism of CNTNAP2. <i>NeuroImage</i> , <b>2010</b> , 53, 1030-42	7.9	89
331	De novo expansion of intermediate alleles in spinocerebellar ataxia 7. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 1809-13	5.6	87
330	The glucocerebrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. <i>Movement Disorders</i> , <b>2013</b> , 28, 232-236	7	86

329	Molecular and clinical study of 18 families with ADCA type II: evidence for genetic heterogeneity and de novo mutation. <i>American Journal of Human Genetics</i> , <b>1999</b> , 64, 1594-603	11	86
328	In vivo skeletal muscle mitochondrial function in Leber's hereditary optic neuropathy assessed by 31P magnetic resonance spectroscopy. <i>Annals of Neurology</i> , <b>1997</b> , 42, 573-9	9.4	81
327	The alpha-synuclein Ala53Thr mutation is not a common cause of familial Parkinson's disease: a study of 230 European cases. European Consortium on Genetic Susceptibility in Parkinson's Disease. <i>Annals of Neurology</i> , <b>1998</b> , 44, 270-3	9.4	81
326	Park6-linked parkinsonism occurs in several european families. <i>Annals of Neurology</i> , <b>2002</b> , 51, 14-18	9.4	81
325	Neuronal intranuclear inclusions in SCA2: a genetic, morphological and immunohistochemical study of two cases. <i>Brain</i> , <b>2002</b> , 125, 656-63	11.2	81
324	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. <i>Movement Disorders</i> , <b>2012</b> , 27, 526-32	7	80
323	The genetics of dystonia: new twists in an old tale. <i>Brain</i> , <b>2013</b> , 136, 2017-37	11.2	80
322	Failure to replicate previously reported genetic associations with sporadic temporal lobe epilepsy: where to from here?. <i>Brain</i> , <b>2005</b> , 128, 1832-40	11.2	79
321	Fine mapping of de novo CMT1A and HNPP rearrangements within CMT1A-REPs evidences two distinct sex-dependent mechanisms and candidate sequences involved in recombination. <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 141-8	5.6	79
320	Polygenic risk of Parkinson disease is correlated with disease age at onset. <i>Annals of Neurology</i> , <b>2015</b> , 77, 582-91	9.4	77
319	A missense mutation in KCTD17 causes autosomal dominant myoclonus-dystonia. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 938-47	11	77
318	Common variants in the HLA-DRB1-HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis. <i>Nature Genetics</i> , <b>2013</b> , 45, 208-13	36.3	76
317	Somatic and germline mosaicism in sporadic early-onset Alzheimer's disease. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 1219-24	5.6	76
316	Dopa-responsive dystonia in British patients: new mutations of the GTP-cyclohydrolase I gene and evidence for genetic heterogeneity. <i>Human Molecular Genetics</i> , <b>1996</b> , 5, 403-6	5.6	76
315	Single-Molecule Imaging of Individual Amyloid Protein Aggregates in Human Biofluids. <i>ACS Chemical Neuroscience</i> , <b>2016</b> , 7, 399-406	5.7	75
314	Bioenergetic consequences of PINK1 mutations in Parkinson disease. <i>PLoS ONE</i> , <b>2011</b> , 6, e25622	3.7	75
313	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 763-71	11	74
312	Mutations in SNX14 cause a distinctive autosomal-recessive cerebellar ataxia and intellectual disability syndrome. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 611-21	11	73

311	Sequencing of the alpha-synuclein gene in a large series of cases of familial Parkinson's disease fails to reveal any further mutations. The European Consortium on Genetic Susceptibility in Parkinson's Disease (GSPD). <i>Human Molecular Genetics</i> , <b>1998</b> , 7, 751-3	5.6	73
310	Genome-wide association studies: the key to unlocking neurodegeneration?. <i>Nature Neuroscience</i> , <b>2010</b> , 13, 789-94	25.5	72
309	Genetic and clinical heterogeneity in paroxysmal kinesigenic dyskinesia: evidence for a third EKD gene. <i>Movement Disorders</i> , <b>2002</b> , 17, 717-25	7	72
308	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , <b>2015</b> , 36, 1605.e7-12	5.6	70
307	Genetic and functional characterisation of the P/Q calcium channel in episodic ataxia with epilepsy. <i>Journal of Physiology</i> , <b>2010</b> , 588, 1905-13	3.9	70
306	Genetic variability at the PARK16 locus. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 1356-9	5.3	69
305	Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. <i>Brain</i> , <b>2020</b> , 143, 234-248	11.2	69
304	Rare individual amyloid- $\beta$ oligomers act on astrocytes to initiate neuronal damage. <i>Biochemistry</i> , <b>2014</b> , 53, 2442-53	3.2	68
303	Hyposmia in G2019S LRRK2-related parkinsonism: clinical and pathologic data. <i>Neurology</i> , <b>2008</b> , 71, 1021-6	16.5	68
302	The phenotypic spectrum of DYT24 due to ANO3 mutations. <i>Movement Disorders</i> , <b>2014</b> , 29, 928-34	7	67
301	Parkin is recruited into aggresomes in a stress-specific manner: over-expression of parkin reduces aggresome formation but can be dissociated from parkin's effect on neuronal survival. <i>Human Molecular Genetics</i> , <b>2004</b> , 13, 117-35	5.6	67
300	The Ile93Met mutation in the ubiquitin carboxy-terminal-hydrolase-L1 gene is not observed in European cases with familial Parkinson's disease. <i>Neuroscience Letters</i> , <b>1999</b> , 270, 1-4	3.3	67
299	The role of the SCA2 trinucleotide repeat expansion in 89 autosomal dominant cerebellar ataxia families. Frequency, clinical and genetic correlates. <i>Brain</i> , <b>1998</b> , 121 ( Pt 3), 459-67	11.2	66
298	Developing and validating Parkinson's disease subtypes and their motor and cognitive progression. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 1279-1287	5.5	66
297	Parkinson's disease is not associated with the combined $\beta$ -synuclein/apolipoprotein E susceptibility genotype. <i>Annals of Neurology</i> , <b>2001</b> , 49, 665-668	9.4	64
296	The genetics of Parkinson's disease. <i>Current Opinion in Genetics and Development</i> , <b>2000</b> , 10, 292-8	4.9	64
295	Loss-of-function mutations in RAB39B are associated with typical early-onset Parkinson disease. <i>Neurology: Genetics</i> , <b>2015</b> , 1, e9	3.8	63
294	ADCY5 mutations are another cause of benign hereditary chorea. <i>Neurology</i> , <b>2015</b> , 85, 80-8	6.5	63

293	Cell death pathways in Parkinson's disease: role of mitochondria. <i>Antioxidants and Redox Signaling</i> , <b>2009</b> , 11, 2135-49	8.4	63
292	A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. <i>Brain</i> , <b>2004</b> , 127, 973-80	11.2	63
291	Localization of the gene for distal hereditary motor neuronopathy VII (dHMN-VII) to chromosome 2q14. <i>American Journal of Human Genetics</i> , <b>2001</b> , 68, 1270-6	11	63
290	Molecular basis of Parkinson's disease. <i>NeuroReport</i> , <b>2009</b> , 20, 150-6	1.7	62
289	Identification and sizing of the GAA trinucleotide repeat expansion of Friedreich's ataxia in 56 patients. Clinical and genetic correlates. <i>Brain</i> , <b>1997</b> , 120 ( Pt 4), 673-80	11.2	60
288	Mutations in HPCA cause autosomal-recessive primary isolated dystonia. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 657-65	11	59
287	Deletions at 22q11.2 in idiopathic Parkinson's disease: a combined analysis of genome-wide association data. <i>Lancet Neurology</i> , <b>2016</b> , 15, 585-96	24.1	59
286	Dopamine induced neurodegeneration in a PINK1 model of Parkinson's disease. <i>PLoS ONE</i> , <b>2012</b> , 7, e37564	3.4	58
285	Multiple sclerosis and the HLA-D region: linkage and association studies. <i>Journal of Neuroimmunology</i> , <b>1995</b> , 58, 183-90	3.5	58
284	Systemic amyloid deposits in familial British dementia. <i>Journal of Biological Chemistry</i> , <b>2001</b> , 276, 43909-14	3.4	57
283	A multicenter study of BRD2 as a risk factor for juvenile myoclonic epilepsy. <i>Epilepsia</i> , <b>2007</b> , 48, 706-12	6.4	56
282	A new family with paroxysmal exercise induced dystonia and migraine: a clinical and genetic study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2000</b> , 68, 609-14	5.5	56
281	Features of -associated Parkinson's disease at presentation in the UK study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 702-709	5.5	55
280	Genotype and phenotype in Parkinson's disease: lessons in heterogeneity from deep brain stimulation. <i>Movement Disorders</i> , <b>2013</b> , 28, 1370-5	7	55
279	Autosomal dominant cerebellar ataxia: SCA2 is the most frequent mutation in eastern India. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2004</b> , 75, 448-52	5.5	55
278	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , <b>2017</b> , 57, 247.e9-247.e13	5.6	54
277	ADCY5-related movement disorders: Frequency, disease course and phenotypic variability in a cohort of paediatric patients. <i>Parkinsonism and Related Disorders</i> , <b>2017</b> , 41, 37-43	3.6	54
276	The correlation between reading and mathematics ability at age twelve has a substantial genetic component. <i>Nature Communications</i> , <b>2014</b> , 5, 4204	17.4	54

275	Glucocerebrosidase mutations in 108 neuropathologically confirmed cases of multiple system atrophy. <i>Neurology</i> , <b>2009</b> , 72, 1185-6	6.5	54
274	Sex-dependent rearrangements resulting in CMT1A and HNPP. <i>Nature Genetics</i> , <b>1997</b> , 17, 136-7	36.3	54
273	Mitochondrial ND5 gene variation associated with encephalomyopathy and mitochondrial ATP consumption. <i>Journal of Biological Chemistry</i> , <b>2007</b> , 282, 36845-52	5.4	53
272	Association between a polymorphism of ubiquitin carboxy-terminal hydrolase L1 (UCH-L1) gene and sporadic Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2000</b> , 6, 195-197	3.6	53
271	Ataxia telangiectasia presenting as dopa-responsive cervical dystonia. <i>Neurology</i> , <b>2013</b> , 81, 1148-51	6.5	52
270	PINK, PANK, or PARK? A clinicians' guide to familial parkinsonism. <i>Lancet Neurology</i> , <b>2004</b> , 3, 652-624.1	24.1	52
269	Unusual phenotypes in DYT1 dystonia: a report of five cases and a review of the literature. <i>Movement Disorders</i> , <b>2003</b> , 18, 706-11	7	51
268	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. <i>JAMA Neurology</i> , <b>2018</b> , 75, 1416-1422	14.2	50
267	Cell metabolism affects selective vulnerability in PINK1-associated Parkinson's disease. <i>Journal of Cell Science</i> , <b>2011</b> , 124, 4194-202	5.3	50
266	Genetic comorbidities in Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 831-41	5.6	49
265	C9ORF72 expansions, parkinsonism, and Parkinson disease: a clinicopathologic study. <i>Neurology</i> , <b>2013</b> , 81, 808-11	6.5	49
264	Dopa-responsive dystonia -- the story so far. <i>Neuropediatrics</i> , <b>2002</b> , 33, 1-5	1.6	49
263	A functional polymorphism regulating dopamine beta-hydroxylase influences against Parkinson's disease. <i>Annals of Neurology</i> , <b>2004</b> , 55, 443-6	9.4	48
262	Targeting mitochondrial dysfunction in neurodegenerative disease: Part I. <i>Expert Opinion on Therapeutic Targets</i> , <b>2010</b> , 14, 369-85	6.4	47
261	Six novel connexin32 (GJB1) mutations in X-linked Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2002</b> , 73, 304-6	5.5	47
260	The pathogenesis of demyelinating disease. <i>Progress in Neurobiology</i> , <b>1994</b> , 43, 143-73	10.9	47
259	Analysis of genome-wide association studies of Alzheimer disease and of Parkinson disease to determine if these 2 diseases share a common genetic risk. <i>JAMA Neurology</i> , <b>2013</b> , 70, 1268-76	17.2	46
258	The role of interruptions in polyQ in the pathology of SCA1. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003648	6	46

257	Connexin 32 promoter P2 mutations: a mechanism of peripheral nerve dysfunction. <i>Annals of Neurology</i> , <b>2004</b> , 56, 730-4	9.4	46
256	Strong association of a novel Tau promoter haplotype in progressive supranuclear palsy. <i>Neuroscience Letters</i> , <b>2001</b> , 311, 145-8	3.3	46
255	Screening for VPS35 mutations in Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 838.e1-5	5.6	45
254	Mitochondrial DNA polymorphisms in pathologically proven Parkinson's disease. <i>Journal of Neurology</i> , <b>1997</b> , 244, 262-5	5.5	45
253	Generalized chorea in two patients harboring the Friedreich's ataxia gene trinucleotide repeat expansion. <i>Movement Disorders</i> , <b>1998</b> , 13, 339-40	7	45
252	Association of MAPT haplotype-tagging SNPs with sporadic Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2009</b> , 30, 1477-82	5.6	44
251	Hereditary sensory neuropathy is caused by a mutation in the delta subunit of the cytosolic chaperonin-containing t-complex peptide-1 (Cct4) gene. <i>Human Molecular Genetics</i> , <b>2003</b> , 12, 1917-25	5.6	44
250	Autosomal recessive, DYT2-like primary torsion dystonia: a new family. <i>Neurology</i> , <b>2003</b> , 61, 1801-3	6.5	44
249	Causes of Parkinson's disease: genetics of DJ-1. <i>Cell and Tissue Research</i> , <b>2004</b> , 318, 185-8	4.2	44
248	CHCHD2 and Parkinson's disease. <i>Lancet Neurology</i> , <b>2015</b> , 14, 678-9	24.1	43
247	Genetic causes of Parkinson's disease: UCHL-1. <i>Cell and Tissue Research</i> , <b>2004</b> , 318, 189-94	4.2	43
246	PREDICT-PD: An online approach to prospectively identify risk indicators of Parkinson's disease. <i>Movement Disorders</i> , <b>2017</b> , 32, 219-226	7	42
245	Parkinsonism and nigrostriatal dysfunction are associated with spinocerebellar ataxia type 6 (SCA6). <i>Movement Disorders</i> , <b>2005</b> , 20, 1115-9	7	42
244	Paroxysmal dystonic choreoathetosis: clinical features and investigation of pathophysiology in a large family. <i>Movement Disorders</i> , <b>2000</b> , 15, 648-57	7	42
243	Cervical dystonia is associated with a polymorphism in the dopamine (D5) receptor gene. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2001</b> , 71, 262-4	5.5	41
242	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , <b>2019</b> , 34, 460-468	7	40
241	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 5483-5489	5.6	40
240	Fine-mapping, gene expression and splicing analysis of the disease associated LRRK2 locus. <i>PLoS ONE</i> , <b>2013</b> , 8, e70724	3.7	40



239	Ataxin-7 aggregation and ubiquitination in infantile SCA7 with 180 CAG repeats. <i>Annals of Neurology</i> , <b>2004</b> , 56, 448-52	9.4	39
238	Autosomal-recessive cerebellar ataxia caused by a novel ADCK3 mutation that elongates the protein: clinical, genetic and biochemical characterisation. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2014</b> , 85, 493-8	5.5	38
237	Neuropathology of primary adult-onset dystonia. <i>Neurology</i> , <b>2008</b> , 70, 695-9	6.5	38
236	The alpha-synuclein gene in multiple system atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2006</b> , 77, 464-7	5.5	38
235	Genome-wide analysis of the parkinsonism-dementia complex of Guam. <i>Archives of Neurology</i> , <b>2004</b> , 61, 1889-97		38
234	SLC25A46 mutations underlie progressive myoclonic ataxia with optic atrophy and neuropathy. <i>Movement Disorders</i> , <b>2016</b> , 31, 1249-51	7	37
233	LRRK2 expression in idiopathic and G2019S positive Parkinson's disease subjects: a morphological and quantitative study. <i>Neuropathology and Applied Neurobiology</i> , <b>2011</b> , 37, 777-90	5.2	37
232	Targeting mitochondrial dysfunction in neurodegenerative disease: Part II. <i>Expert Opinion on Therapeutic Targets</i> , <b>2010</b> , 14, 497-511	6.4	37
231	Dejerine-Sottas neuropathy and PMP22 point mutations: a new base pair substitution and a possible "hot spot" on Ser72. <i>Annals of Neurology</i> , <b>1998</b> , 43, 680-3	9.4	37
230	Synphilin-1 and parkin show overlapping expression patterns in human brain and form aggresomes in response to proteasomal inhibition. <i>Neurobiology of Disease</i> , <b>2005</b> , 20, 401-11	7.5	37
229	LRRK2 activation controls the repair of damaged endomembranes in macrophages. <i>EMBO Journal</i> , <b>2020</b> , 39, e104494	13	37
228	A genome-wide association analysis of a broad psychosis phenotype identifies three loci for further investigation. <i>Biological Psychiatry</i> , <b>2014</b> , 75, 386-97	7.9	36
227	Nigrostriatal dysfunction in homozygous and heterozygous parkin gene carriers: an 18F-dopa PET progression study. <i>Movement Disorders</i> , <b>2009</b> , 24, 2260-6	7	36
226	Genome-wide scan linkage analysis for Parkinson's disease: the European genetic study of Parkinson's disease. <i>Journal of Medical Genetics</i> , <b>2004</b> , 41, 900-7	5.8	36
225	Equating scores of the University of Pennsylvania Smell Identification Test and Sniffin' Sticks test in patients with Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2016</b> , 33, 96-101	3.6	36
224	Genetic analysis of Mendelian mutations in a large UK population-based Parkinson's disease study. <i>Brain</i> , <b>2019</b> , 142, 2828-2844	11.2	35
223	Genetic aspects of Parkinson's disease. <i>Movement Disorders</i> , <b>1998</b> , 13, 203-11	7	35
222	The tau locus is not significantly associated with pathologically confirmed sporadic Parkinson's disease. <i>Neuroscience Letters</i> , <b>2002</b> , 330, 201-3	3.3	35



221	Hyperexcitable substantia nigra dopamine neurons in PINK1- and HtrA2/Omi-deficient mice. <i>Journal of Neurophysiology</i> , <b>2010</b> , 104, 3009-20	3.2	34
220	Differential DJ-1 gene expression in Parkinson's disease. <i>Neurobiology of Disease</i> , <b>2009</b> , 36, 393-400	7.5	34
219	Mouse models for neurological disease. <i>Lancet Neurology, The</i> , <b>2002</b> , 1, 215-24	24.1	34
218	Familial adult onset of Krabbe's disease resembling hereditary spastic paraplegia with normal neuroimaging. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2002</b> , 72, 635-8	5.5	34
217	Neurofibrillary tangle parkinsonian disorders--tau pathology and tau genetics. <i>Movement Disorders</i> , <b>1999</b> , 14, 731-6	7	34
216	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. <i>New England Journal of Medicine</i> , <b>2021</b> , 385, 1868-1880	59.2	34
215	Study of the genetic variability in a Parkinson's Disease gene: EIF4G1. <i>Neuroscience Letters</i> , <b>2012</b> , 518, 19-22	3.3	33
214	Loss-of-Function Variants in HOPS Complex Genes VPS16 and VPS41 Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , <b>2020</b> , 88, 867-877	9.4	33
213	PINK1 deficiency in $\beta$ cells increases basal insulin secretion and improves glucose tolerance in mice. <i>Open Biology</i> , <b>2014</b> , 4, 140051	7	32
212	Myoclonus-dystonia syndrome due to tyrosine hydroxylase deficiency. <i>Neurology</i> , <b>2012</b> , 79, 435-41	6.5	32
211	The gene responsible for PARK6 Parkinson's disease, PINK1, does not influence common forms of parkinsonism. <i>Annals of Neurology</i> , <b>2004</b> , 56, 329-35	9.4	32
210	The sepiapterin reductase gene region reveals association in the PARK3 locus: analysis of familial and sporadic Parkinson's disease in European populations. <i>Journal of Medical Genetics</i> , <b>2006</b> , 43, 557-62	5.8	32
209	Detailed genotyping demonstrates association between the slow acetylator genotype for N-acetyltransferase 2 (NAT2) and familial Parkinson's disease. <i>Movement Disorders</i> , <b>2000</b> , 15, 30-5	7	32
208	Tracking Parkinson's: Study Design and Baseline Patient Data. <i>Journal of Parkinson's Disease</i> , <b>2015</b> , 5, 947-59	5.3	31
207	Characterisation of a novel NR4A2 mutation in Parkinson's disease brain. <i>Neuroscience Letters</i> , <b>2009</b> , 457, 75-9	3.3	31
206	Partial epilepsy with pericentral spikes: a new familial epilepsy syndrome with evidence for linkage to chromosome 4p15. <i>Annals of Neurology</i> , <b>2002</b> , 51, 740-9	9.4	31
205	Immunological study of hereditary motor and sensory neuropathy type 1a (HMSN1a). <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2002</b> , 72, 230-5	5.5	31
204	Polymorphism in a lincRNA Associates with a Doubled Risk of Pneumococcal Bacteremia in Kenyan Children. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 1092-1100	11	30

203	HtrA2 deficiency causes mitochondrial uncoupling through the F <sub>1</sub> F <sub>0</sub> ATP synthase and consequent ATP depletion. <i>Cell Death and Disease</i> , <b>2012</b> , 3, e335	9.8	30
202	Molecular genetic pathways in Parkinson's disease: a review. <i>Clinical Science</i> , <b>2005</b> , 109, 355-64	6.5	30
201	Phenotypic variation of a new P0 mutation in genetically identical twins. <i>Journal of Neurology</i> , <b>1999</b> , 246, 596-9	5.5	30
200	Susceptibility to multiple sclerosis and the immunoglobulin heavy chain variable region. <i>Journal of Neurology</i> , <b>1995</b> , 242, 677-82	5.5	30
199	Multiple sclerosis in the Cambridge health district of east Anglia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>1992</b> , 55, 877-82	5.5	30
198	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations. <i>Journal of Allergy and Clinical Immunology</i> , <b>2020</b> , 146, 901-911	11.5	29
197	Mendelian randomization study shows no causal relationship between circulating urate levels and Parkinson's disease. <i>Annals of Neurology</i> , <b>2018</b> , 84, 191-199	9.4	29
196	The Parkinson's Disease Mendelian Randomization Research Portal. <i>Movement Disorders</i> , <b>2019</b> , 34, 1864-1872		29
195	Cathepsin D deficiency causes juvenile-onset ataxia and distinctive muscle pathology. <i>Neurology</i> , <b>2014</b> , 83, 1873-5	6.5	29
194	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. <i>Neurology</i> , <b>2012</b> , 79, 127-31	6.5	29
193	Detection of novel mutations and review of published data suggests that hereditary spastic paraplegia caused by spastin (SPAST) mutations is found more often in males. <i>Journal of the Neurological Sciences</i> , <b>2011</b> , 306, 62-5	3.2	29
192	Paroxysmal dystonic choreoathetosis. Genetic linkage studies in a British family. <i>Brain</i> , <b>1997</b> , 120 (Pt 12), 2125-30	11.2	29
191	Large-scale pathways-based association study in amyotrophic lateral sclerosis. <i>Brain</i> , <b>2007</b> , 130, 2292-301	11.2	29
190	Genetics of movement disorders and ataxia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2002</b> , 73 Suppl 2, II22-6	5.5	29
189	Use of support vector machines for disease risk prediction in genome-wide association studies: concerns and opportunities. <i>Human Mutation</i> , <b>2012</b> , 33, 1708-18	4.7	28
188	Phosphorylation of HtrA2 by cyclin-dependent kinase-5 is important for mitochondrial function. <i>Cell Death and Differentiation</i> , <b>2012</b> , 19, 257-66	12.7	28
187	Alternative ion channel splicing in mesial temporal lobe epilepsy and Alzheimer's disease. <i>Genome Biology</i> , <b>2007</b> , 8, R32	18.3	28
186	Slowly progressive cerebellar ataxia and cervical dystonia: clinical presentation of a new form of spinocerebellar ataxia?. <i>Movement Disorders</i> , <b>2003</b> , 18, 200-6	7	28

185	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , <b>2020</b> , 35, 774-780	7	27
184	The genetic associations of acute anterior uveitis and their overlap with the genetics of ankylosing spondylitis. <i>Genes and Immunity</i> , <b>2016</b> , 17, 46-51	4.4	27
183	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. <i>Human Mutation</i> , <b>2018</b> , 39, 965-969	4.7	27
182	No major role of common SV2A variation for predisposition or levetiracetam response in epilepsy. <i>Epilepsy Research</i> , <b>2009</b> , 83, 44-51	3	27
181	Nonmotor symptoms in Parkin gene-related parkinsonism. <i>Movement Disorders</i> , <b>2010</b> , 25, 1279-84	7	27
180	Depletion of mitochondrial DNA by ddC in untransformed human cell lines. <i>Somatic Cell and Molecular Genetics</i> , <b>1997</b> , 23, 287-90		27
179	Familial dopa-responsive cervical dystonia. <i>Neurology</i> , <b>2006</b> , 66, 599-601	6.5	27
178	PARK6 is a common cause of familial parkinsonism. <i>Neurological Sciences</i> , <b>2002</b> , 23 Suppl 2, S117-8	3.5	27
177	Genome-Wide Association Studies of Cognitive and Motor Progression in Parkinson's Disease. <i>Movement Disorders</i> , <b>2021</b> , 36, 424-433	7	27
176	Effects of age and MAOA genotype on the neural processing of social rejection. <i>Genes, Brain and Behavior</i> , <b>2010</b> , 9, 628-37	3.6	26
175	GJB1 gene mutations in suspected inflammatory demyelinating neuropathies not responding to treatment. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2009</b> , 80, 699-700	5.5	26
174	Parkin disease in a Brazilian kindred: Manifesting heterozygotes and clinical follow-up over 10 years. <i>Movement Disorders</i> , <b>2005</b> , 20, 479-484	7	26
173	Chromosome 13 dementia syndromes as models of neurodegeneration. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , <b>2001</b> , 8, 277-84	2.7	26
172	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 969-977	11	25
171	Typical features of Parkinson disease and diagnostic challenges with microdeletion 22q11.2. <i>Neurology</i> , <b>2018</b> , 90, e2059-e2067	6.5	25
170	Susceptibility loci for pigmentation and melanoma in relation to Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2014</b> , 35, 1512.e5-1512.e10	5.6	25
169	Clinical heterogeneity and genotype-phenotype correlations in hereditary spastic paraplegia because of Spatacsin mutations (SPG11). <i>European Journal of Neurology</i> , <b>2008</b> , 15, 1065-70	6	25
168	Motor cortical physiology in patients and asymptomatic carriers of parkin gene mutations. <i>Movement Disorders</i> , <b>2008</b> , 23, 1812-9	7	25

167	Apolipoprotein E4 is probably responsible for the chromosome 19 linkage peak for Parkinson's disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2005</b> , 136B, 72-4	3.5	25
166	Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 4653-60	5.6	24
165	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2018</b> , 64, 159.e5-159.e8	5.6	23
164	Tremor-ataxia with central hypomyelination (TACH): dystonia as a new clinical feature. <i>Movement Disorders</i> , <b>2012</b> , 27, 1829-30	7	23
163	Expression of BRI2 mRNA and protein in normal human brain and familial British dementia: its relevance to the pathogenesis of disease. <i>Neuropathology and Applied Neurobiology</i> , <b>2008</b> , 34, 492-505	5.2	23
162	Corticobasal degeneration syndrome with basal ganglia calcification: Fahr's disease as a corticobasal look-alike?. <i>Movement Disorders</i> , <b>2002</b> , 17, 563-7	7	23
161	No linkage or association between multiple sclerosis and the myelin basic protein gene in affected sibling pairs. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>1994</b> , 57, 1191-4	5.5	23
160	In vivo assessment of brain monoamine systems in parkin gene carriers: a PET study. <i>Experimental Neurology</i> , <b>2010</b> , 222, 120-4	5.7	22
159	Mitochondria in Parkinson disease: back in fashion with a little help from genetics. <i>Archives of Neurology</i> , <b>2006</b> , 63, 649-54		22
158	A pharmacogenetic exploration of vigabatrin-induced visual field constriction. <i>Epilepsy Research</i> , <b>2006</b> , 70, 144-52	3	22
157	What have PINK1 and HtrA2 genes told us about the role of mitochondria in Parkinson's disease?. <i>Annals of the New York Academy of Sciences</i> , <b>2008</b> , 1147, 30-6	6.5	21
156	PARK11 is not linked with Parkinson's disease in European families. <i>European Journal of Human Genetics</i> , <b>2005</b> , 13, 193-7	5.3	21
155	Effect of ApoE and tau on age of onset of progressive supranuclear palsy and multiple system atrophy. <i>Neuroscience Letters</i> , <b>2001</b> , 312, 118-20	3.3	21
154	Heterogeneity in clinical features and disease severity in ataxia-associated SYNE1 mutations. <i>Journal of Neurology</i> , <b>2016</b> , 263, 1503-10	5.5	21
153	Olfaction in Parkin single and compound heterozygotes in a cohort of young onset Parkinson's disease patients. <i>Acta Neurologica Scandinavica</i> , <b>2016</b> , 134, 271-6	3.8	20
152	The role of the mitochondrial NCX in the mechanism of neurodegeneration in Parkinson's disease. <i>Advances in Experimental Medicine and Biology</i> , <b>2013</b> , 961, 241-9	3.6	20
151	Nonsyndromic Parkinson disease in a family with autosomal dominant optic atrophy due to mutations. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e188	3.8	20
150	Tau acts as an independent genetic risk factor in pathologically proven PD. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 838.e7-11	5.6	20

149	The human homologue of the weaver mouse gene in familial and sporadic Parkinson's disease. <i>Neuroscience</i> , <b>1996</b> , 72, 877-9	3.9	20
148	Dystonia genes functionally converge in specific neurons and share neurobiology with psychiatric disorders. <i>Brain</i> , <b>2020</b> , 143, 2771-2787	11.2	20
147	The CACNA1B R1389H variant is not associated with myoclonus-dystonia in a large European multicentric cohort. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 5326-9	5.6	19
146	ALS2 mutations: juvenile amyotrophic lateral sclerosis and generalized dystonia. <i>Neurology</i> , <b>2014</b> , 82, 1065-7	6.5	19
145	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. <i>Nature Genetics</i> , <b>2021</b> , 53, 1636-1648	36.3	19
144	Statins are underused in recent-onset Parkinson's disease with increased vascular risk: findings from the UK Tracking Parkinson's and Oxford Parkinson's Disease Centre (OPDC) discovery cohorts. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2016</b> , 87, 1183-1190	5.5	19
143	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , <b>2019</b> , 34, 1851-1863	7.6	18
142	PDXK mutations cause polyneuropathy responsive to pyridoxal 5'-phosphate supplementation. <i>Annals of Neurology</i> , <b>2019</b> , 86, 225-240	9.4	18
141	Analysis of Parkinson's disease brain-derived DNA for alpha-synuclein coding somatic mutations. <i>Movement Disorders</i> , <b>2014</b> , 29, 1060-4	7	18
140	Autonomic Dysfunction in Early Parkinson's Disease: Results from the United Kingdom Tracking Parkinson's Study. <i>Movement Disorders Clinical Practice</i> , <b>2017</b> , 4, 509-516	2.2	18
139	Atypical parkinsonism with apraxia and supranuclear gaze abnormalities in type 1 Gaucher disease. Expanding the spectrum: case report and literature review. <i>Movement Disorders</i> , <b>2010</b> , 25, 1506-9	7	18
138	Prader-Willi and Angelman syndromes: update on genetic mechanisms and diagnostic complexities. <i>Current Opinion in Neurology</i> , <b>1999</b> , 12, 149-54	7.1	18
137	The GTP-cyclohydrolase I gene in atypical parkinsonian patients: a clinico-genetic study. <i>Journal of the Neurological Sciences</i> , <b>1996</b> , 141, 27-32	3.2	18
136	Truncating mutations in patients are associated with a high rate of psychiatric comorbidities in hereditary spastic paraplegia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2017</b> , 88, 681-687	5.5	18
135	Cooperative genome-wide analysis shows increased homozygosity in early onset Parkinson's disease. <i>PLoS ONE</i> , <b>2012</b> , 7, e28787	3.7	18
134	Neuronal intranuclear inclusion disease is genetically heterogeneous. <i>Annals of Clinical and Translational Neurology</i> , <b>2020</b> , 7, 1716-1725	5.3	18
133	Variation in Recent Onset Parkinson's Disease: Implications for Prodromal Detection. <i>Journal of Parkinson's Disease</i> , <b>2016</b> , 6, 289-300	5.3	18
132	PDE10A and ADCY5 mutations linked to molecular and microstructural basal ganglia pathology. <i>Movement Disorders</i> , <b>2018</b> , 33, 1961-1965	7	18

131	Genotype-phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. <i>Movement Disorders</i> , <b>2018</b> , 33, 1119-1129	7	17
130	Clinical and genetic analysis of spinocerebellar ataxia type 11. <i>Cerebellum</i> , <b>2008</b> , 7, 159-64	4.3	17
129	Nigral degeneration and striatal dopaminergic dysfunction in idiopathic and Parkin-linked Parkinson's disease. <i>Movement Disorders</i> , <b>2006</b> , 21, 299-305	7	17
128	No pathogenic mutations in the synphilin-1 gene in Parkinson's disease. <i>Neuroscience Letters</i> , <b>2001</b> , 307, 125-7	3.3	17
127	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , <b>2021</b> , 78, 464-472	17.2	17
126	Stratification of candidate genes for Parkinson's disease using weighted protein-protein interaction network analysis. <i>BMC Genomics</i> , <b>2018</b> , 19, 452	4.5	17
125	Genetic association studies of complex neurological diseases. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2006</b> , 77, 1302-4	5.5	16
124	Examining the role of common genetic variation in the gamma2 subunit of the GABA(A) receptor in epilepsy using tagging SNPs. <i>Epilepsy Research</i> , <b>2006</b> , 70, 229-38	3	16
123	Reduction in endogenous parkin levels renders glial cells sensitive to both caspase-dependent and caspase-independent cell death. <i>European Journal of Neuroscience</i> , <b>2004</b> , 20, 2038-48	3.5	16
122	Mitochondrial DNA point mutation T9176C in Leigh syndrome. <i>Journal of Child Neurology</i> , <b>2000</b> , 15, 830-3.5		16
121	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 1851-1862	8.1	16
120	Migraine with aura as the predominant phenotype in a family with a PRRT2 mutation. <i>Journal of Neurology</i> , <b>2013</b> , 260, 656-60	5.5	15
119	Genetic screening of Greek patients with Huntington's disease phenocopies identifies an SCA8 expansion. <i>Journal of Neurology</i> , <b>2012</b> , 259, 1874-8	5.5	15
118	The syndrome of (predominantly cervical) dystonia and cerebellar ataxia: new cases indicate a distinct but heterogeneous entity. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2007</b> , 78, 774-5	5.5	15
117	UCHL-1 gene in multiple system atrophy: a haplotype tagging approach. <i>Movement Disorders</i> , <b>2005</b> , 20, 1338-43	7	15
116	Identification of a novel primary torsion dystonia locus (DYT13) on chromosome 1p36 in an Italian family with cranial-cervical or upper limb onset. <i>Neurological Sciences</i> , <b>2001</b> , 22, 95-6	3.5	15
115	Intrafamilial phenotypic variability in Friedreich ataxia associated with a G130V mutation in the FRDA gene. <i>Archives of Neurology</i> , <b>2002</b> , 59, 296-300		15
114	Genetic risk factors in Parkinson's disease. <i>Annals of Neurology</i> , <b>1998</b> , 44, S58-62	9.4	15



113	Mutations in XRCC1 cause cerebellar ataxia and peripheral neuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 1230-1232	5.5	14
112	Is the MC1R variant p.R160W associated with Parkinson's?. <i>Annals of Neurology</i> , <b>2016</b> , 79, 159-61	9.4	14
111	Evidence for pre and postsynaptic nigrostriatal dysfunction in the fragile X tremor-ataxia syndrome. <i>Movement Disorders</i> , <b>2009</b> , 24, 1245-7	7	14
110	NR4A2 genetic variation in sporadic Parkinson's disease: a genome-wide approach. <i>Movement Disorders</i> , <b>2006</b> , 21, 1960-3	7	14
109	Mutations in the gene encoding human peryn are not associated with amyotrophic lateral sclerosis or familial Parkinson's disease. <i>Neuroscience Letters</i> , <b>1999</b> , 274, 21-4	3.3	14
108	The genetics of Parkinson's disease. <i>Current Opinion in Neurology</i> , <b>1999</b> , 12, 427-32	7.1	14
107	LRP10 in Synucleinopathies. <i>Lancet Neurology</i> , <b>2018</b> , 17, 1032	24.1	14
106	RFC1 Intronic Repeat Expansions Absent in Pathologically Confirmed Multiple Systems Atrophy. <i>Movement Disorders</i> , <b>2020</b> , 35, 1277-1279	7	13
105	Polygenic risk of ischemic stroke is associated with cognitive ability. <i>Neurology</i> , <b>2016</b> , 86, 611-8	6.5	13
104	Assessment of Parkinson's disease risk loci in Greece. <i>Neurobiology of Aging</i> , <b>2014</b> , 35, 442.e9-442.e16	5.6	12
103	The frequency of spinocerebellar ataxia type 23 in a UK population. <i>Journal of Neurology</i> , <b>2013</b> , 260, 856-9	5.5	12
102	Genetic and Clinical Heterogeneity in Thirteen New Cases with Aceruloplasminemia. Atypical Anemia as a Clue for an Early Diagnosis. <i>International Journal of Molecular Sciences</i> , <b>2020</b> , 21,	6.3	11
101	Establishing the role of rare coding variants in known Parkinson's disease risk loci. <i>Neurobiology of Aging</i> , <b>2017</b> , 59, 220.e11-220.e18	5.6	11
100	Preliminary investigation of the influence of dopamine regulating genes on social working memory. <i>Social Neuroscience</i> , <b>2014</b> , 9, 437-51	2	11
99	Endothelial, sympathetic, and cardiac function in inherited (6R)-L-erythro-5,6,7,8-tetrahydro-L-biopterin deficiency. <i>Circulation: Cardiovascular Genetics</i> , <b>2010</b> , 3, 513-22		11
98	Mutation analysis of the sodium/hydrogen exchanger gene (NHE5) in familial paroxysmal kinesigenic dyskinesia. <i>Journal of Neural Transmission</i> , <b>2002</b> , 109, 1189-94	4.3	11
97	No linkage between multiple sclerosis and the T cell receptor alpha chain locus. <i>Journal of the Neurological Sciences</i> , <b>1994</b> , 124, 32-7	3.2	11
96	Late-onset Lafora disease with prominent parkinsonism due to a rare mutation in EPM2A. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e101	3.8	11



95	Low Prevalence of NOTCH2NLC GGC Repeat Expansion in White Patients with Movement Disorders. <i>Movement Disorders</i> , <b>2021</b> , 36, 251-255	7	11
94	-related ataxia is a mimic of early multiple system atrophy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2021</b> ,	5.5	11
93	Complexity of the Genetics and Clinical Presentation of Spinocerebellar Ataxia 17. <i>Frontiers in Cellular Neuroscience</i> , <b>2018</b> , 12, 429	6.1	11
92	Genome-Wide Association Study Identifies Risk Loci for Cluster Headache. <i>Annals of Neurology</i> , <b>2021</b> , 90, 193-202	9.4	11
91	L-dopa responsiveness in early Parkinson's disease is associated with the rate of motor progression. <i>Parkinsonism and Related Disorders</i> , <b>2019</b> , 65, 55-61	3.6	10
90	Influence of COMT genotype and affective distractors on the processing of self-generated thought. <i>Social Cognitive and Affective Neuroscience</i> , <b>2015</b> , 10, 777-82	4	10
89	Advances in the Genetics of Parkinson's Disease: A Guide for the Clinician. <i>Movement Disorders Clinical Practice</i> , <b>2014</b> , 1, 3-13	2.2	10
88	Mutational analysis of parkin and PINK1 in multiple system atrophy. <i>Neurobiology of Aging</i> , <b>2011</b> , 32, 548.e5-7	5.6	10
87	Multiple mitochondrial DNA deletions in monozygotic twins with OPMD. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2008</b> , 79, 68-71	5.5	10
86	Population genetics for target identification. <i>Drug Discovery Today: Technologies</i> , <b>2004</b> , 1, 69-74	7.1	10
85	Sequence analysis of tau in familial and sporadic progressive supranuclear palsy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2002</b> , 72, 388-90	5.5	10
84	Utility of the new Movement Disorder Society clinical diagnostic criteria for Parkinson's disease applied retrospectively in a large cohort study of recent onset cases. <i>Parkinsonism and Related Disorders</i> , <b>2017</b> , 40, 40-46	3.6	9
83	GGC repeat expansion in NOTCH2NLC is rare in European patients with essential tremor. <i>Brain</i> , <b>2020</b> , 143, e57	11.2	9
82	Genetic variation in is associated with bacteremia secondary to diverse pathogens in African children. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2018</b> , 115, E3601-E3603	11.5	9
81	An intragenic duplication in guanosine triphosphate cyclohydrolase-1 gene in a dopa-responsive dystonia family. <i>Movement Disorders</i> , <b>2011</b> , 26, 905-9	7	9
80	Novel peripheral myelin protein 22 (PMP22) micromutations associated with variable phenotypes in Greek patients with Charcot-Marie-Tooth disease. <i>Brain</i> , <b>2012</b> , 135, e217, 1-6; author reply e218, 1-2	11.2	9
79	Analysis of DNM3 and VAMP4 as genetic modifiers of LRRK2 Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2021</b> , 97, 148.e17-148.e24	5.6	9
78	LRP10 in Synucleinopathies. <i>Lancet Neurology, The</i> , <b>2018</b> , 17, 1033-1034	24.1	9

77	loss of function causes autosomal recessive spastic ataxia and optic atrophy. <i>Annals of Clinical and Translational Neurology</i> , <b>2018</b> , 5, 216-221	5.3	8
76	Characterisation and validation of insertions and deletions in 173 patient exomes. <i>PLoS ONE</i> , <b>2012</b> , 7, e51292	3.7	8
75	Dissecting the Phenotype and Genotype of PLA2G6-Related Parkinsonism. <i>Movement Disorders</i> , <b>2021</b> ,	7	8
74	-related disease is associated with central pontine calcifications and atypical parkinsonism. <i>Neurology: Genetics</i> , <b>2020</b> , 6, e399	3.8	7
73	Friedreich's ataxia and other hereditary ataxias in Greece: an 18-year perspective. <i>Journal of the Neurological Sciences</i> , <b>2014</b> , 336, 87-92	3.2	7
72	No pathogenic GNAL mutations in 192 sporadic and familial cases of cervical dystonia. <i>Movement Disorders</i> , <b>2014</b> , 29, 154-5	7	7
71	Mutation of the sterol 27-hydroxylase gene ( CYP27A1) in a Taiwanese family with cerebrotendinous xanthomatosis. <i>Journal of Neurology</i> , <b>2002</b> , 249, 1311-2	5.5	7
70	DNA repair in trinucleotide repeat ataxias. <i>FEBS Journal</i> , <b>2018</b> , 285, 3669-3682	5.7	7
69	The ADH1C stop mutation in multiple system atrophy patients and healthy probands in the United Kingdom and Germany. <i>Movement Disorders</i> , <b>2006</b> , 21, 2034	7	6
68	Association of genetic loci: replication or not, that is the question. <i>Neurology</i> , <b>2005</b> , 64, 1989	6.5	6
67	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , <b>2021</b> , 90, 35-42	9.4	6
66	GGC Repeat Expansion in NOTCH2NLC Is Rare in European Leukoencephalopathy. <i>Annals of Neurology</i> , <b>2020</b> , 88, 641-642	9.4	5
65	Diagnostic clues and manifesting carriers in fukutin-related protein (FKRP) limb-girdle muscular dystrophy. <i>Journal of the Neurological Sciences</i> , <b>2015</b> , 348, 266-8	3.2	5
64	Analysis of spinocerebellar ataxias due to expanded triplet repeats in Greek patients with cerebellar ataxia. <i>Journal of the Neurological Sciences</i> , <b>2012</b> , 318, 178-80	3.2	5
63	Identification of UBAP1 mutations in juvenile hereditary spastic paraplegia in the 100,000 Genomes Project. <i>European Journal of Human Genetics</i> , <b>2020</b> , 28, 1763-1768	5.3	5
62	Delineating the phenotype of autosomal-recessive HPCA mutations: Not only isolated dystonia!. <i>Movement Disorders</i> , <b>2019</b> , 34, 589-592	7	4
61	Reply: Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , <b>2015</b> , 138, e352	11.2	4
60	Sequencing analysis of the SCA6 CAG expansion excludes an influence of repeat interruptions on disease onset. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 1226-1227	5.5	4

59	Running a neurogenetic clinic. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2002</b> , 73 Suppl 2, II2-4	5.5	4
58	Integration of eQTL and Parkinson disease GWAS data implicates 11 disease genes		4
57	Using Mendelian randomization to understand and develop treatments for neurodegenerative disease. <i>Brain Communications</i> , <b>2020</b> , 2, fcaa031	4.5	4
56	Expanding the Spectrum of AP5Z1-Related Hereditary Spastic Paraplegia (HSP-SPG48): A Multicenter Study on a Rare Disease. <i>Movement Disorders</i> , <b>2021</b> , 36, 1034-1038	7	4
55	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 562-562	5.6	3
54	Population genetic approaches to neurological disease: Parkinson's disease as an example. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , <b>2005</b> , 360, 1573-8	5.8	3
53	Mitochondrial disorders in neuro-ophthalmology. <i>Current Opinion in Neurology</i> , <b>1996</b> , 9, 1-4	7.1	3
52	The Parkinson Disease Mendelian Randomization Research Portal		3
51	Ca <sup>2+</sup> is a key factor in $\alpha$ -synuclein-induced neurotoxicity. <i>Development (Cambridge)</i> , <b>2016</b> , 143, e1.1-e1.1	6.6	3
50	Mitochondrial DNA Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. <i>Annals of Neurology</i> , <b>2021</b> , 89, 1240-1247	9.4	3
49	Identification of sixteen novel candidate genes for late onset Parkinson's disease. <i>Molecular Neurodegeneration</i> , <b>2021</b> , 16, 35	19	3
48	NOTCH2NLC Intermediate-Length Repeat Expansion and Parkinson's Disease in Patients of European Descent. <i>Annals of Neurology</i> , <b>2021</b> , 89, 633-635	9.4	3
47	Variant Alzheimer's disease with spastic paraparesis and cotton wool plaques is caused by PS-1 mutations that lead to exceptionally high amyloid- $\beta$ concentrations <b>2000</b> , 48, 806		3
46	DYT13, a novel primary torsion dystonia locus, maps to chromosome 1p36.13B6.32 in an Italian family with cranial-cervical or upper limb onset <b>2001</b> , 49, 362		3
45	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , <b>2019</b> , 180, 223-231	3.5	2
44	The repeat variant in MSH3 is not a genetic modifier for spinocerebellar ataxia type 3 and Friedreich's ataxia. <i>Brain</i> , <b>2020</b> , 143, e25	11.2	2
43	The influence of microsatellite polymorphisms in sex steroid receptor genes ESR1, ESR2 and AR on sex differences in brain structure. <i>NeuroImage</i> , <b>2020</b> , 221, 117087	7.9	2
42	Mutational analysis of PMP22, EGR2, LITAF and NEFL in Greek Charcot-Marie-Tooth type 1 patients. <i>Clinical Genetics</i> , <b>2013</b> , 83, 388-91	4	2

41	Assessment of a DJ-1 (PARK7) polymorphism in Finnish PD. <i>Neurology</i> , <b>2004</b> , 62, 2335	6.5	2
40	The paroxysmal dyskinesias <b>2001</b> , 125-140		2
39	Neurogenetics: A Guide for Clinicians <b>2012</b> ,		2
38	Regulation of mitophagy by the NSL complex underlies genetic risk for Parkinson's disease at Chr16q11.2 and on the MAPT H1 allele		2
37	Biallelic variants in TSPOAP1, encoding the active-zone protein RIMBP1, cause autosomal recessive dystonia. <i>Journal of Clinical Investigation</i> , <b>2021</b> , 131,	15.9	2
36	Childhood-Onset Chorea Caused by a Recurrent De Novo DRD2 Variant. <i>Movement Disorders</i> , <b>2021</b> , 36, 1472-1473	7	2
35	Triple trouble: a striking new phenotype or competing genes in a family with hereditary spastic paraplegia. <i>Journal of Neurology</i> , <b>2016</b> , 263, 1232-3	5.5	2
34	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. <i>Nature Communications</i> , <b>2021</b> , 12, 7342	17.4	2
33	Screening of mutations in NOL3 in a myoclonic syndromes series. <i>Journal of Neurology</i> , <b>2014</b> , 261, 1830-3	5.5	1
32	Hypersomnia with dilated pupils in adenosine monophosphate deaminase (AMPD) deficiency. <i>Journal of Sleep Research</i> , <b>2014</b> , 23, 118-20	5.8	1
31	Genetic approaches to solving common diseases. <i>Journal of Neurology</i> , <b>2004</b> , 251, 1169-72	5.5	1
30	Introduction: genetic variation and human health. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , <b>2005</b> , 360, 1539-41	5.8	1
29	Cerebellar and Midbrain Lysosomal Enzyme Deficiency in Isolated Dystonia.. <i>Movement Disorders</i> , <b>2022</b> ,	7	1
28	Bi-allelic variants in TSPOAP1, encoding the active zone protein RIMBP1, cause autosomal recessive dystonia		1
27	Mendelian Randomisation Finds No Causal Association between Urate and Parkinson's Disease Progression. <i>Movement Disorders</i> , <b>2021</b> , 36, 2182-2187	7	1
26	Spastic paraplegia preceding -related familial Alzheimer's disease. <i>Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring</i> , <b>2021</b> , 13, e12186	5.2	1
25	The mitochondrial DNA G13513A transition in ND5 is associated with a LHON/MELAS overlap syndrome and may be a frequent cause of MELAS <b>1999</b> , 46, 916		1
24	Dopa-Responsive Parkinsonism in a Patient With Homozygous RFC1 Expansions. <i>Movement Disorders</i> , <b>2020</b> , 35, 1890-1891	7	0

23	Fibrillation and molecular characteristics are coherent with clinical and pathological features of 4-repeat tauopathy caused by MAPT variant G273R. <i>Neurobiology of Disease</i> , <b>2020</b> , 146, 105079	7.5	o
22	The role of body fat in multiple sclerosis susceptibility and severity: A Mendelian randomisation study.. <i>Multiple Sclerosis Journal</i> , <b>2022</b> , 13524585221092644	5	o
21	Cerebellar Ataxias and Related Conditions <b>2016</b> , 685-698		
20	EFFECTS OF VASCULAR COMORBIDITY IN PARKINSON'S DISEASE. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2016</b> , 87, e1.13-e1	5.5	
19	When the penny drops. <i>Practical Neurology</i> , <b>2014</b> , 14, 409-14	2.4	
18	Genetic linkage analysis of a large family with photoparoxysmal response. <i>Epilepsy Research</i> , <b>2012</b> , 99, 38-45	3	
17	The ataxias52-63		
16	Parkinsonism83-102		
15	Channelopathies121-135		
14	FAMILY HISTORY IN YOUNG ONSET PARKINSON'S DISEASE. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2013</b> , 84, e2.69-e2	5.5	
13	TRACKING PARKINSON'S (THE PROBAND STUDY)INTERIM REPORT FROM THE FIRST 1000 CASES. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2013</b> , 84, e2.70-e2	5.5	
12	Interview: The genetics of Parkinson disease: piecing together the jigsaw. <i>Neurodegenerative Disease Management</i> , <b>2011</b> , 1, 105-107	2.8	
11	Ataxia in a young patient. <i>Practical Neurology</i> , <b>2011</b> , 11, 319-22	2.4	
10	Cerebellar Ataxias and Related Conditions629-643		
9	Progressive cognitive decline with truncal/limb ataxia and ballistic movements. <i>Movement Disorders</i> , <b>1997</b> , 12, 1075-84	7	
8	Genetic testing in neurology. <i>Medicine</i> , <b>2008</b> , 36, 566-568	0.6	
7	Genetics of progressive supranuclear palsy. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn</i> , <b>2008</b> , 89, 475-85	3	
6	Commentary on "A genome wide linkage disequilibrium screen in Parkinson's disease" by Foltynie et al. in <i>J Neurol</i> (2005) 252:597-602. <i>Journal of Neurology</i> , <b>2005</b> , 252, 603-4	5.5	

5 Pharmacogenomics and the Treatment of Neurological Disease 337-346

4 Genetics of the overlap between epilepsy and movement disorders **2001**, 451-464

3 Mutations in Nuclear Genes That Affect Mitochondrial Function in Parkinson Disease **2012**, 43-61

2 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

1 Clinical and genetic analysis of spinocerebellar ataxia type 11. *Cerebellum*, **2008**, 7, 1-6 4-3