

# Matthew Farrer

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

343  
papers

29,855  
citations

77  
h-index

165  
g-index

356  
ext. papers

33,444  
ext. citations

7.4  
avg, IF

6.5  
L-index

#	Paper	IF	Citations
343	Dairy Intake and Parkinson's Disease: A Mendelian Randomization Study.. <i>Movement Disorders</i> , <b>2022</b> ,	7	2
342	The Gut-Brain Axis and Its Relation to Parkinson's Disease: A Review.. <i>Frontiers in Aging Neuroscience</i> , <b>2021</b> , 13, 782082	5.3	5
341	Mendelian Randomisation Study of Smoking, Alcohol, and Coffee Drinking in Relation to Parkinson's Disease. <i>Journal of Parkinsons Disease</i> , <b>2021</b> ,	5.3	3
340	Hunting for Familial Parkinson's Disease Mutations in the Post Genome Era. <i>Genes</i> , <b>2021</b> , 12,	4.2	1
339	Dynamic control of the dopamine transporter in neurotransmission and homeostasis. <i>Npj Parkinsons Disease</i> , <b>2021</b> , 7, 22	9.7	14
338	Reply: UQCRC1 variants in Parkinson's disease: a large cohort study in Chinese mainland population. <i>Brain</i> , <b>2021</b> , 144, e55	11.2	
337	Genomewide Association Studies of LRRK2 Modifiers of Parkinson's Disease. <i>Annals of Neurology</i> , <b>2021</b> , 90, 76-88	9.4	9
336	LRRK2; a dynamic regulator of cellular trafficking. <i>Brain Research</i> , <b>2021</b> , 1761, 147394	3.7	0
335	-associated neurodevelopmental disorder. <i>Journal of Medical Genetics</i> , <b>2021</b> , 58, 196-204	5.8	4
334	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
333	Chronic and Acute Manipulation of Cortical Glutamate Transmission Induces Structural and Synaptic Changes in Co-cultured Striatal Neurons. <i>Frontiers in Cellular Neuroscience</i> , <b>2021</b> , 15, 569031	6.1	0
332	Parkinson disease risk variants in East Asian populations. <i>Nature Reviews Neurology</i> , <b>2020</b> , 16, 461-462	15	2
331	Neuron-autonomous susceptibility to induced synuclein aggregation is exacerbated by endogenous mutations and ameliorated by genetic knock-out. <i>Brain Communications</i> , <b>2020</b> , 2, fcz052	4.5	8
330	Variants in saposin D domain of prosaposin gene linked to Parkinson's disease. <i>Brain</i> , <b>2020</b> , 143, 1190-1205	15.2	37
329	Disease modification and biomarker development in Parkinson disease: Revision or reconstruction?. <i>Neurology</i> , <b>2020</b> , 94, 481-494	6.5	60
328	Neuropathological findings in PINK1-associated Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2020</b> , 78, 105-108	3.6	7
327	Nonsteroidal Anti-inflammatory Use and LRRK2 Parkinson's Disease Penetrance. <i>Movement Disorders</i> , <b>2020</b> , 35, 1755-1764	7	21

326	Mitochondrial UQCRC1 mutations cause autosomal dominant parkinsonism with polyneuropathy. <i>Brain</i> , <b>2020</b> , 143, 3352-3373	11.2	16
325	Age at Onset of LRRK2 p.Gly2019Ser Is Related to Environmental and Lifestyle Factors. <i>Movement Disorders</i> , <b>2020</b> , 35, 1854-1858	7	7
324	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit-successes and challenges. <i>European Journal of Pediatrics</i> , <b>2019</b> , 178, 1207-1218	4.1	29
323	Mitochondrial DNA Deletions Discriminate Affected from Unaffected LRRK2 Mutation Carriers. <i>Annals of Neurology</i> , <b>2019</b> , 86, 324-326	9.4	8
322	DNAJC13 p.Asn855Ser, implicated in familial parkinsonism, alters membrane dynamics of sorting nexin 1. <i>Neuroscience Letters</i> , <b>2019</b> , 706, 114-122	3.3	5
321	Doubts about TMEM230 as a gene for parkinsonism. <i>Nature Genetics</i> , <b>2019</b> , 51, 367-368	36.3	8
320	Whole-Exome Sequencing of an Exceptional Longevity Cohort. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , <b>2019</b> , 74, 1386-1390	6.4	9
319	Family with primary periodic paralysis and a mutation in MCM3AP, a gene implicated in mRNA transport. <i>Muscle and Nerve</i> , <b>2019</b> , 60, 311-314	3.4	2
318	Diagnostic Yield and Treatment Impact of Targeted Exome Sequencing in Early-Onset Epilepsy. <i>Frontiers in Neurology</i> , <b>2019</b> , 10, 434	4.1	42
317	Pathophysiology of and therapeutic options for a GABRA1 variant linked to epileptic encephalopathy. <i>Molecular Brain</i> , <b>2019</b> , 12, 92	4.5	4
316	Single Inflammatory Trigger Leads to Neuroinflammation in LRRK2 Rodent Model without Degeneration of Dopaminergic Neurons. <i>Journal of Parkinsons Disease</i> , <b>2019</b> , 9, 121-139	5.3	9
315	Pipeline to gene discovery - Analysing familial Parkinsonism in the Queensland Parkinson's Project. <i>Parkinsonism and Related Disorders</i> , <b>2018</b> , 49, 34-41	3.6	11
314	The emerging role of Rab GTPases in the pathogenesis of Parkinson's disease. <i>Movement Disorders</i> , <b>2018</b> , 33, 196-207	7	29
313	Altered dopamine release and monoamine transporters in Vps35 p.D620N knock-in mice. <i>Npj Parkinsons Disease</i> , <b>2018</b> , 4, 27	9.7	38
312	Assessing an Interactive Online Tool to Support Parents' Genomic Testing Decisions. <i>Journal of Genetic Counseling</i> , <b>2018</b> , 28, 10	2.5	13
311	p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. <i>Journal of Movement Disorders</i> , <b>2018</b> , 11, 45-48	2.9	10
310	Establishing diagnostic criteria for Perry syndrome. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2018</b> , 89, 482-487	5.5	26
309	Dopamine receptors and BDNF-haplotypes predict dyskinesia in Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2018</b> , 47, 39-44	3.6	25

308	A Meta-Analysis of $\beta$ -Synuclein Multiplication in Familial Parkinsonism. <i>Frontiers in Neurology</i> , <b>2018</b> , 9, 1021	4.1	46
307	A Case of Parkinson's Disease with No Lewy Body Pathology due to a Homozygous Exon Deletion in. <i>Case Reports in Neurological Medicine</i> , <b>2018</b> , 2018, 6838965	0.7	24
306	Age- and disease-dependent increase of the mitophagy marker phospho-ubiquitin in normal aging and Lewy body disease. <i>Autophagy</i> , <b>2018</b> , 14, 1404-1418	10.2	47
305	Gender differences in Parkinson's disease depression. <i>Parkinsonism and Related Disorders</i> , <b>2017</b> , 36, 93-97	3.6	21
304	Genetic Identification in Early Onset Parkinsonism among Norwegian Patients. <i>Movement Disorders Clinical Practice</i> , <b>2017</b> , 4, 499-508	2.2	13
303	SCA2 family presenting as typical Parkinson's disease: 34 year follow up. <i>Parkinsonism and Related Disorders</i> , <b>2017</b> , 40, 69-72	3.6	8
302	An Infant With Epilepsy and Recurrent Hemiplegia due to Compound Heterozygous Variants in ATP1A2. <i>Pediatric Neurology</i> , <b>2017</b> , 75, 87-90	2.9	12
301	Reply: Heterozygous PINK1 p.G411S in rapid eye movement sleep behaviour disorder. <i>Brain</i> , <b>2017</b> , 140, e33	11.2	2
300	Neurobehavioral characterization of adult-onset Alexander disease: A family study. <i>Neurology: Clinical Practice</i> , <b>2017</b> , 7, 425-429	1.7	3
299	Serotonin and dopamine transporter PET changes in the premotor phase of LRRK2 parkinsonism: cross-sectional studies. <i>Lancet Neurology</i> , <b>2017</b> , 16, 351-359	24.1	64
298	Initial elevations in glutamate and dopamine neurotransmission decline with age, as does exploratory behavior, in LRRK2 G2019S knock-in mice. <i>ELife</i> , <b>2017</b> , 6,	8.9	44
297	DNAJC12 and dopa-responsive nonprogressive parkinsonism. <i>Annals of Neurology</i> , <b>2017</b> , 82, 640-646	9.4	38
296	De Novo Mutations in YWHAG Cause Early-Onset Epilepsy. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 300-310	11	37
295	Homozygous alpha-synuclein p.A53V in familial Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2017</b> , 57, 248.e7-248.e12	5.6	51
294	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 65-74	11	58
293	Heterozygous PINK1 p.G411S increases risk of Parkinson's disease via a dominant-negative mechanism. <i>Brain</i> , <b>2017</b> , 140, 98-117	11.2	88
292	Evaluation of the interaction between LRRK2 and PARK16 loci in determining risk of Parkinson's disease: analysis of a large multicenter study. <i>Neurobiology of Aging</i> , <b>2017</b> , 49, 217.e1-217.e4	5.6	5
291	De novo mutation in 2 patients with neonatal-onset epilepsy. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e120	3.8	18

290	Leucine-rich repeat kinase 2 is a regulator of B cell function, affecting homeostasis, BCR signaling, IgA production, and T1 antigen responses. <i>Journal of Neuroimmunology</i> , <b>2016</b> , 292, 1-8	3.5	8
289	Lovastatin protects neurite degeneration in LRRK2-G2019S parkinsonism through activating the Akt/Nrf pathway and inhibiting GSK3 $\beta$ activity. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 1965-1978	5.6	34
288	$\beta$ synuclein genetic variability: A biomarker for dementia in Parkinson disease. <i>Annals of Neurology</i> , <b>2016</b> , 79, 991-9	9.4	64
287	A scan without evidence is not evidence of absence: Scans without evidence of dopaminergic deficit in a symptomatic leucine-rich repeat kinase 2 mutation carrier. <i>Movement Disorders</i> , <b>2016</b> , 31, 405-9	7	12
286	Double homozygous mutations (R275W and M432V) in the ParkinGene associated with late-onset Parkinson's disease. <i>Movement Disorders</i> , <b>2016</b> , 31, 423-5	7	2
285	Conjugal parkinsonism is coincidental. <i>Parkinsonism and Related Disorders</i> , <b>2016</b> , 33, 149-150	3.6	2
284	Leucine-rich repeat kinase 2 (LRRK2) regulates $\beta$ synuclein clearance in microglia. <i>BMC Neuroscience</i> , <b>2016</b> , 17, 77	3.2	37
283	DCTN1 p.K56R in progressive supranuclear palsy. <i>Parkinsonism and Related Disorders</i> , <b>2016</b> , 28, 56-61	3.6	24
282	DNM3 and genetic modifiers of age of onset in LRRK2 Gly2019Ser parkinsonism: a genome-wide linkage and association study. <i>Lancet Neurology, The</i> , <b>2016</b> , 15, 1248-1256	24.1	50
281	Conjugal parkinsonism - Clinical, pathology and genetic study. No evidence of person-to-person transmission. <i>Parkinsonism and Related Disorders</i> , <b>2016</b> , 31, 87-90	3.6	11
280	DNAJC13 genetic variants in parkinsonism. <i>Movement Disorders</i> , <b>2015</b> , 30, 273-8	7	32
279	Novel LRRK2 mutations in Parkinsonism. <i>Parkinsonism and Related Disorders</i> , <b>2015</b> , 21, 1119-21	3.6	6
278	Defining neurodegeneration on Guam by targeted genomic sequencing. <i>Annals of Neurology</i> , <b>2015</b> , 77, 458-68	9.4	47
277	Progressive dopaminergic alterations and mitochondrial abnormalities in LRRK2 G2019S knock-in mice. <i>Neurobiology of Disease</i> , <b>2015</b> , 78, 172-95	7.5	140
276	[11C]PBR28 PET imaging is sensitive to neuroinflammation in the aged rat. <i>Journal of Cerebral Blood Flow and Metabolism</i> , <b>2015</b> , 35, 1331-8	7.3	21
275	Insights from late-onset familial parkinsonism on the pathogenesis of idiopathic Parkinson's disease. <i>Lancet Neurology, The</i> , <b>2015</b> , 14, 1054-64	24.1	45
274	Large-scale assessment of polyglutamine repeat expansions in Parkinson disease. <i>Neurology</i> , <b>2015</b> , 85, 1283-92	6.5	20
273	Chronic and acute LRRK2 silencing has no long-term behavioral effects, whereas wild-type and mutant LRRK2 overexpression induce motor and cognitive deficits and altered regulation of dopamine release. <i>Parkinsonism and Related Disorders</i> , <b>2015</b> , 21, 1156-63	3.6	30

272	Retromer-dependent neurotransmitter receptor trafficking to synapses is altered by the Parkinson's disease VPS35 mutation p.D620N. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1691-703	5.6	98
271	Parkinson's disease, genetic variability and the Faroe Islands. <i>Parkinsonism and Related Disorders</i> , <b>2015</b> , 21, 75-8	3.6	9
270	Parkinsonism in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , <b>2015</b> , 138, e349	11.2	17
269	Head injury, $\alpha$ -Synuclein genetic variability and Parkinson's disease. <i>European Journal of Neurology</i> , <b>2015</b> , 22, 874-8	6	12
268	LRRK2 overexpression alters glutamatergic presynaptic plasticity, striatal dopamine tone, postsynaptic signal transduction, motor activity and memory. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1336-49	5.6	70
267	Genetic variability of the retromer cargo recognition complex in parkinsonism. <i>Movement Disorders</i> , <b>2015</b> , 30, 580-4	7	20
266	Familial aggregation of Parkinson's disease in the Faroe Islands. <i>Movement Disorders</i> , <b>2015</b> , 30, 538-44	7	13
265	DNAJC13 p.Asn855Ser mutation screening in Parkinson's disease and pathologically confirmed Lewy body disease patients. <i>European Journal of Neurology</i> , <b>2015</b> , 22, 1323-5	6	15
264	Motor phenotype of LRRK2-associated Parkinson's disease: a Tunisian longitudinal study. <i>Movement Disorders</i> , <b>2015</b> , 30, 253-8	7	24
263	A novel DCTN1 mutation with late-onset parkinsonism and frontotemporal atrophy. <i>Movement Disorders</i> , <b>2014</b> , 29, 1201-4	7	30
262	DNAJC13 mutations in Parkinson disease. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 1794-801	5.6	209
261	Protective effect of LRRK2 p.R1398H on risk of Parkinson's disease is independent of MAPT and SNCA variants. <i>Neurobiology of Aging</i> , <b>2014</b> , 35, 266.e5-14	5.6	26
260	Identification of FUS p.R377W in essential tremor. <i>European Journal of Neurology</i> , <b>2014</b> , 21, 361-3	6	23
259	LRRK2 parkinsonism in Tunisia and Norway: a comparative analysis of disease penetrance. <i>Neurology</i> , <b>2014</b> , 83, 568-9	6.5	32
258	Does $\alpha$ -Synuclein have a dual and opposing effect in preclinical vs. clinical Parkinson's disease?. <i>Parkinsonism and Related Disorders</i> , <b>2014</b> , 20, 584-9; discussion 584	3.6	31
257	EIF4G1 gene mutations are not a common cause of Parkinson's disease in the Japanese population. <i>Parkinsonism and Related Disorders</i> , <b>2014</b> , 20, 659-61	3.6	10
256	Clinical, positron emission tomography, and pathological studies of DNAJC13 p.N855S Parkinsonism. <i>Movement Disorders</i> , <b>2014</b> , 29, 1684-7	7	15
255	Synaptic function is modulated by LRRK2 and glutamate release is increased in cortical neurons of G2019S LRRK2 knock-in mice. <i>Frontiers in Cellular Neuroscience</i> , <b>2014</b> , 8, 301	6.1	72

254	Disease penetrance of late-onset parkinsonism: a meta-analysis. <i>JAMA Neurology</i> , <b>2014</b> , 71, 1535-9	17.2	65
253	LRRK2 exonic variants and risk of multiple system atrophy. <i>Neurology</i> , <b>2014</b> , 83, 2256-61	6.5	34
252	In vivo dopaminergic and serotonergic dysfunction in DCTN1 gene mutation carriers. <i>Movement Disorders</i> , <b>2014</b> , 29, 1197-201	7	12
251	Michael J. Fox Foundation LRRK2 Consortium: geographical differences in returning genetic research data to study participants. <i>Genetics in Medicine</i> , <b>2014</b> , 16, 644-5	8.1	7
250	The role of SNCA and MAPT in Parkinson disease and LRRK2 parkinsonism in the Tunisian Arab-Berber population. <i>European Journal of Neurology</i> , <b>2014</b> , 21, e91-2	6	6
249	Behavioral deficits and striatal DA signaling in LRRK2 p.G2019S transgenic rats: a multimodal investigation including PET neuroimaging. <i>Journal of Parkinsons Disease</i> , <b>2014</b> , 4, 483-98	5.3	27
248	Genetics and genomics of Parkinson's disease. <i>Genome Medicine</i> , <b>2014</b> , 6, 48	14.4	125
247	Comparative study of Parkinson's disease and leucine-rich repeat kinase 2 p.G2019S parkinsonism. <i>Neurobiology of Aging</i> , <b>2014</b> , 35, 1125-31	5.6	64
246	Measurements of Dopaminergic Function in the Rat Brain Using [18F]FDOPA PET and Microdialysis <b>2014</b> , 161		
245	Advances in the genetics of Parkinson disease. <i>Nature Reviews Neurology</i> , <b>2013</b> , 9, 445-54	15	353
244	The genetics of Parkinson's disease: progress and therapeutic implications. <i>Movement Disorders</i> , <b>2013</b> , 28, 14-23	7	248
243	Population-specific frequencies for LRRK2 susceptibility variants in the Genetic Epidemiology of Parkinson's Disease (GEO-PD) Consortium. <i>Movement Disorders</i> , <b>2013</b> , 28, 1740-4	7	24
242	LRRK2 phosphorylates novel tau epitopes and promotes tauopathy. <i>Acta Neuropathologica</i> , <b>2013</b> , 126, 809-27	14.3	67
241	STX6 rs1411478 is not associated with increased risk of Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2013</b> , 19, 563-5	3.6	14
240	Sequence variants in eukaryotic translation initiation factor 4-gamma (eIF4G1) are associated with Lewy body dementia. <i>Acta Neuropathologica</i> , <b>2013</b> , 125, 425-38	14.3	16
239	Measuring dopaminergic function in the 6-OHDA-lesioned rat: a comparison of PET and microdialysis. <i>EJNMMI Research</i> , <b>2013</b> , 3, 69	3.6	14
238	Alpha-synuclein p.H50Q, a novel pathogenic mutation for Parkinson's disease. <i>Movement Disorders</i> , <b>2013</b> , 28, 811-3	7	433
237	In-vivo measurement of LDOPA uptake, dopamine reserve and turnover in the rat brain using [18F]FDOPA PET. <i>Journal of Cerebral Blood Flow and Metabolism</i> , <b>2013</b> , 33, 59-66	7.3	21

236	Patient-control association study of the Leucine-Rich repeat kinase 2 (LRRK2) gene in South African Parkinson's disease patients. <i>Movement Disorders</i> , <b>2013</b> , 28, 2039-40	7	4
235	A multi-centre clinico-genetic analysis of the VPS35 gene in Parkinson disease indicates reduced penetrance for disease-associated variants. <i>Journal of Medical Genetics</i> , <b>2012</b> , 49, 721-6	5.8	78
234	Cognitive dysfunction in Tunisian LRRK2 associated Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , <b>2012</b> , 18, 243-6	3.6	31
233	First neuropathological description of a patient with Parkinson's disease and LRRK2 p.N1437H mutation. <i>Parkinsonism and Related Disorders</i> , <b>2012</b> , 18, 332-8	3.6	31
232	PARK2 variability in Polish Parkinson's disease patients--interaction with mitochondrial haplogroups. <i>Parkinsonism and Related Disorders</i> , <b>2012</b> , 18, 520-4	3.6	9
231	Large-scale replication and heterogeneity in Parkinson disease genetic loci. <i>Neurology</i> , <b>2012</b> , 79, 659-67	6.5	106
230	Polymorphic genes of detoxification and mitochondrial enzymes and risk for progressive supranuclear palsy: a case control study. <i>BMC Medical Genetics</i> , <b>2012</b> , 13, 16	2.1	3
229	LRRK2 knockout mice have an intact dopaminergic system but display alterations in exploratory and motor co-ordination behaviors. <i>Molecular Neurodegeneration</i> , <b>2012</b> , 7, 25	19	139
228	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
227	An evaluation of the impact of MAPT, SNCA and APOE on the burden of Alzheimer's and Lewy body pathology. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2012</b> , 83, 424-9	5.5	46
226	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. <i>Nature Genetics</i> , <b>2011</b> , 43, 699-705	36.3	386
225	Glucocerebrosidase mutations in diffuse Lewy body disease. <i>Parkinsonism and Related Disorders</i> , <b>2011</b> , 17, 55-7	3.6	37
224	Subclinical signs in LRRK2 mutation carriers. <i>Parkinsonism and Related Disorders</i> , <b>2011</b> , 17, 528-32	3.6	30
223	Lrrk2 p.Q1111H substitution and Parkinson's disease in Latin America. <i>Parkinsonism and Related Disorders</i> , <b>2011</b> , 17, 629-31	3.6	12
222	A large-scale genetic association study to evaluate the contribution of Omi/HtrA2 (PARK13) to Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2011</b> , 32, 548.e9-18	5.6	46
221	Role of sepiapterin reductase gene at the PARK3 locus in Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2011</b> , 32, 2108.e1-5	5.6	15
220	Death-associated protein kinase 1 variation and Parkinson's disease. <i>European Journal of Neurology</i> , <b>2011</b> , 18, 1090-3	6	4
219	SNCA, MAPT, and GSK3B in Parkinson disease: a gene-gene interaction study. <i>European Journal of Neurology</i> , <b>2011</b> , 18, 876-81	6	31



218	Parkinson-related genetics in patients treated with deep brain stimulation. <i>Acta Neurologica Scandinavica</i> , <b>2011</b> , 123, 201-6	3.8	23
217	Adult neurogenesis and neurite outgrowth are impaired in LRRK2 G2019S mice. <i>Neurobiology of Disease</i> , <b>2011</b> , 41, 706-16	7.5	146
216	Association of LRRK2 exonic variants with susceptibility to Parkinson's disease: a case-control study. <i>Lancet Neurology, The</i> , <b>2011</b> , 10, 898-908	24.1	237
215	VPS35 mutations in Parkinson disease. <i>American Journal of Human Genetics</i> , <b>2011</b> , 89, 162-7	11	618
214	VPS35 Mutations in Parkinson Disease. <i>American Journal of Human Genetics</i> , <b>2011</b> , 89, 347	11	3
213	Translation initiator EIF4G1 mutations in familial Parkinson disease. <i>American Journal of Human Genetics</i> , <b>2011</b> , 89, 398-406	11	213
212	Call for participation in the neurogenetics consortium within the Human Variome Project. <i>Neurogenetics</i> , <b>2011</b> , 12, 169-73	3	4
211	Common variants in PARK loci and related genes and Parkinson's disease. <i>Movement Disorders</i> , <b>2011</b> , 26, 280-8	7	28
210	Genetic variants of Synuclein are not associated with essential tremor. <i>Movement Disorders</i> , <b>2011</b> , 26, 2552-6	7	12
209	Independent and joint effects of the MAPT and SNCA genes in Parkinson disease. <i>Annals of Neurology</i> , <b>2011</b> , 69, 778-92	9.4	76
208	Mutations in LRRK2 increase phosphorylation of peroxiredoxin 3 exacerbating oxidative stress-induced neuronal death. <i>Human Mutation</i> , <b>2011</b> , 32, 1390-7	4.7	80
207	Functional alteration of PARL contributes to mitochondrial dysregulation in Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 1966-74	5.6	131
206	Genome-wide association study identifies novel restless legs syndrome susceptibility loci on 2p14 and 16q12.1. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1002171	6	135
205	A family with Parkinsonism, essential tremor, restless legs syndrome, and depression. <i>Neurology</i> , <b>2011</b> , 76, 1623-30	6.5	25
204	Missing pieces in the Parkinson's disease puzzle. <i>Nature Medicine</i> , <b>2010</b> , 16, 653-61	50.5	521
203	Alpha-synuclein suppression by targeted small interfering RNA in the primate substantia nigra. <i>PLoS ONE</i> , <b>2010</b> , 5, e12122	3.7	101
202	Association of alpha-, beta-, and gamma-Synuclein with diffuse lewy body disease. <i>Archives of Neurology</i> , <b>2010</b> , 67, 970-5		49
201	LRRK2 and Parkinson disease. <i>Archives of Neurology</i> , <b>2010</b> , 67, 542-7		114

200	βSynuclein gene may interact with environmental factors in increasing risk of Parkinson's disease. <i>Neuroepidemiology</i> , <b>2010</b> , 35, 191-5	5.4	52
199	Elucidating the genetics and pathology of Perry syndrome. <i>Journal of the Neurological Sciences</i> , <b>2010</b> , 289, 149-54	3.2	35
198	Glucocerebrosidase mutations are not a common risk factor for Parkinson disease in North Africa. <i>Neuroscience Letters</i> , <b>2010</b> , 477, 57-60	3.3	16
197	Mitochondrial translation initiation factor 3 polymorphism and Parkinson's disease. <i>Neuroscience Letters</i> , <b>2010</b> , 486, 228-30	3.3	17
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182	Comprehensive sequencing of the LRRK2 gene in patients with familial Parkinson's disease from North Africa. <i>Movement Disorders</i> , <b>2010</b> , 25, 2052-8	7	13
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40	Identification and functional characterization of a novel R621C mutation in the synphilin-1 gene in Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2003</b> , 12, 1223-31	5.6	110
39	RING finger 1 mutations in Parkin produce altered localization of the protein. <i>Human Molecular Genetics</i> , <b>2003</b> , 12, 2957-65	5.6	111

38	Clinical, 18F-dopa PET, and genetic analysis of an ethnic Chinese kindred with early-onset parkinsonism and parkin gene mutations. <i>Movement Disorders</i> , <b>2002</b> , 17, 670-5	7	42
37	Case-control study of dopamine transporter-1, monoamine oxidase-B, and catechol-O-methyl transferase polymorphisms in Parkinson's disease. <i>Movement Disorders</i> , <b>2002</b> , 17, 1305-11	7	41
36	Case-control study of estrogen receptor gene polymorphisms in Parkinson's disease. <i>Movement Disorders</i> , <b>2002</b> , 17, 509-12	7	13
35	Accurate determination of ataxin-2 polyglutamine expansion in patients with intermediate-range repeats. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2002</b> , 6, 217-20		15
34	Functional association of the parkin gene promoter with idiopathic Parkinson's disease. <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 2787-92	5.6	81
33	Parkin protects against the toxicity associated with mutant alpha-synuclein: proteasome dysfunction selectively affects catecholaminergic neurons. <i>Neuron</i> , <b>2002</b> , 36, 1007-19	13.9	506
32	A family with a tau P301L mutation presenting with parkinsonism. <i>Parkinsonism and Related Disorders</i> , <b>2002</b> , 9, 121-3	3.6	11
31	Parkinson's genetics: molecular insights for the new millennium. <i>NeuroToxicology</i> , <b>2002</b> , 23, 503-14	4.4	17
30	Tau neurotoxicity without the lesions: a fly challenges a tangled web. <i>Trends in Neurosciences</i> , <b>2002</b> , 25, 327-9	13.3	21
29	The human sideroflexin 5 (SFXN5) gene: sequence, expression analysis and exclusion as a candidate for PARK3. <i>Gene</i> , <b>2002</b> , 285, 229-37	3.8	18
28	Identification and characterization of the human parkin gene promoter. <i>Journal of Neurochemistry</i> , <b>2001</b> , 78, 1146-52	6	25
27	Case-Control study of the extended tau gene haplotype in Parkinson's disease. <i>Annals of Neurology</i> , <b>2001</b> , 50, 658-61	9.4	48
26	Refinement of the PARK3 locus on chromosome 2p13 and the analysis of 14 candidate genes. <i>European Journal of Human Genetics</i> , <b>2001</b> , 9, 659-66	5.3	42
25	Spinocerebellar ataxia type 3 phenotypically resembling parkinson disease in a black family. <i>Archives of Neurology</i> , <b>2001</b> , 58, 296-9		117
24	Genetic analysis of synphilin-1 in familial Parkinson's disease. <i>Neurobiology of Disease</i> , <b>2001</b> , 8, 317-23	7.5	17
23	Lack of nigral pathology in transgenic mice expressing human alpha-synuclein driven by the tyrosine hydroxylase promoter. <i>Neurobiology of Disease</i> , <b>2001</b> , 8, 535-9	7.5	242
22	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
21	Transfected synphilin-1 forms cytoplasmic inclusions in HEK293 cells. <i>Molecular Brain Research</i> , <b>2001</b> , 97, 94-102		53

20	Sensitization of neuronal cells to oxidative stress with mutated human alpha-synuclein. <i>Journal of Neurochemistry</i> , <b>2000</b> , 75, 2546-54	6	71
19	Case-control study of debrisoquine 4-hydroxylase, N-acetyltransferase 2, and apolipoprotein E gene polymorphisms in Parkinson's disease. <i>Movement Disorders</i> , <b>2000</b> , 15, 714-9	7	41
18	Heterotrismy, a significant contributing factor to ventricular septal defect associated with Down syndrome?. <i>Human Genetics</i> , <b>2000</b> , 107, 476-82	6.3	22
17	alpha-Synuclein shares physical and functional homology with 14-3-3 proteins. <i>Journal of Neuroscience</i> , <b>1999</b> , 19, 5782-91	6.6	469
16	A chromosome 4p haplotype segregating with Parkinson's disease and postural tremor. <i>Human Molecular Genetics</i> , <b>1999</b> , 8, 81-5	5.6	177
15	The genetics of disorders with synuclein pathology and parkinsonism. <i>Human Molecular Genetics</i> , <b>1999</b> , 8, 1901-5	5.6	27
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12	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
11	A variant of Alzheimer's disease with spastic paraparesis and unusual plaques due to deletion of exon 9 of presenilin 1. <i>Nature Medicine</i> , <b>1998</b> , 4, 452-5	50.5	302
10	Molecular mapping of Alzheimer-type dementia in Down's syndrome. <i>Annals of Neurology</i> , <b>1998</b> , 43, 380-3	34	275
9	Low frequency of alpha-synuclein mutations in familial Parkinson's disease. <i>Annals of Neurology</i> , <b>1998</b> , 43, 394-7	9.4	134
8	Allelic variability in D21S11, but not in APP or APOE, is associated with cognitive decline in Down syndrome. <i>NeuroReport</i> , <b>1997</b> , 8, 1645-9	1.7	18
7	Localization of frontotemporal dementia with parkinsonism in an Australian kindred to chromosome 17q21-22. <i>Annals of Neurology</i> , <b>1997</b> , 42, 794-8	9.4	76
6	Genetic variation in the COL6A1 region is associated with congenital heart defects in trisomy 21 (Down's syndrome). <i>Annals of Human Genetics</i> , <b>1995</b> , 59, 253-69	2.2	46
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4	The SON gene encodes a conserved DNA binding protein mapping to human chromosome 21. <i>Annals of Human Genetics</i> , <b>1994</b> , 58, 25-34	2.2	24
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2	TMEM230 is not a gene for Parkinson's disease	2
1	Diagnostic Yield and Treatment Impact of Targeted Exome Sequencing in Early-onset Epilepsy	2