

# Andrew Singleton

## List of Publications by Year in descending order

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164  
papers

28,952  
citations

12330

69  
h-index

5539

163  
g-index

185  
all docs

185  
docs citations

185  
times ranked

37026  
citing authors

#	ARTICLE	IF	CITATIONS
1	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. <i>Neuron</i> , 2011, 72, 257-268.	8.1	3,833
2	<i>TREM2</i> Variants in Alzheimer's Disease. <i>New England Journal of Medicine</i> , 2013, 368, 117-127.	27.0	2,385
3	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. <i>Nature Genetics</i> , 2010, 42, 105-116.	21.4	1,982
4	Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. <i>Nature</i> , 2011, 478, 103-109.	27.8	1,855
5	The Parkinson Progression Marker Initiative (PPMI). <i>Progress in Neurobiology</i> , 2011, 95, 629-635.	5.7	1,278
6	Common genetic variants influence human subcortical brain structures. <i>Nature</i> , 2015, 520, 224-229.	27.8	772
7	Kinase activity is required for the toxic effects of mutant LRRK2/dardarin. <i>Neurobiology of Disease</i> , 2006, 23, 329-341.	4.4	683
8	Genomewide Association Studies and Human Disease. <i>New England Journal of Medicine</i> , 2009, 360, 1759-1768.	27.0	683
9	Glucocerebrosidase mutations in clinical and pathologically proven Parkinson's disease. <i>Brain</i> , 2009, 132, 1783-1794.	7.6	612
10	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. <i>Nature Genetics</i> , 2010, 42, 142-148.	21.4	591
11	Lewy bodies and parkinsonism in families with parkin mutations. <i>Annals of Neurology</i> , 2001, 50, 293-300.	5.3	479
12	A Genome-Wide Association Study Identifies Protein Quantitative Trait Loci (pQTLs). <i>PLoS Genetics</i> , 2008, 4, e1000072.	3.5	415
13	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. <i>Nature Genetics</i> , 2011, 43, 1005-1011.	21.4	403
14	Population-Based Genome-wide Association Studies Reveal Six Loci Influencing Plasma Levels of Liver Enzymes. <i>American Journal of Human Genetics</i> , 2008, 83, 520-528.	6.2	402
15	Characterization of PLA2G6 as a locus for dystonia and parkinsonism. <i>Annals of Neurology</i> , 2009, 65, 19-23.	5.3	399
16	A common LRRK2 mutation in idiopathic Parkinson's disease. <i>Lancet</i> , 2005, 365, 415-416.	18.7	391
17	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology</i> , 2006, 5, 911-916.	10.2	360
18	Genome-Wide Association Study of Plasma Polyunsaturated Fatty Acids in the InCHIANTI Study. <i>PLoS Genetics</i> , 2009, 5, e1000338.	3.5	351

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19	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016, 98, 500-513.	6.2	333
20	The Parkinson's progression markers initiative (PPMI) – establishing a PD biomarker cohort. <i>Annals of Clinical and Translational Neurology</i> , 2018, 5, 1460-1477.	3.7	330
21	Association of Cerebrospinal Fluid $\beta$ -Amyloid 1-42, T-tau, P-tau <sub>181</sub> , and $\alpha$ -Synuclein Levels With Clinical Features of Drug-Naive Patients With Early Parkinson Disease. <i>JAMA Neurology</i> , 2013, 70, 1277-87.	9.0	318
22	Using Exome Sequencing to Reveal Mutations in TREM2 Presenting as a Frontotemporal Dementia-like Syndrome Without Bone Involvement. <i>JAMA Neurology</i> , 2013, 70, 78.	9.0	311
23	Genetics of Parkinson's disease and parkinsonism. <i>Annals of Neurology</i> , 2006, 60, 389-398.	5.3	281
24	GWAS of Longevity in CHARGE Consortium Confirms APOE and FOXO3 Candidacy. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2015, 70, 110-118.	3.6	250
25	Novel genetic loci associated with hippocampal volume. <i>Nature Communications</i> , 2017, 8, 13624.	12.8	250
26	Genetic screening for a single common LRRK2 mutation in familial Parkinson's disease. <i>Lancet</i> , The, 2005, 365, 410-412.	13.7	243
27	Early-onset Parkinson's disease caused by a compound heterozygous DJ-1 mutation. <i>Annals of Neurology</i> , 2003, 54, 271-274.	5.3	233
28	Human aging is characterized by focused changes in gene expression and deregulation of alternative splicing. <i>Aging Cell</i> , 2011, 10, 868-878.	6.7	230
29	Genome-wide Association Study of Vitamin B6, Vitamin B12, Folate, and Homocysteine Blood Concentrations. <i>American Journal of Human Genetics</i> , 2009, 84, 477-482.	6.2	225
30	Novel genetic loci underlying human intracranial volume identified through genome-wide association. <i>Nature Neuroscience</i> , 2016, 19, 1569-1582.	14.8	213
31	Genome-wide SNP assay reveals structural genomic variation, extended homozygosity and cell-line induced alterations in normal individuals. <i>Human Molecular Genetics</i> , 2007, 16, 1-14.	2.9	211
32	Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology</i> , The, 2007, 6, 322-328.	10.2	206
33	A Nonsense Mutation in COQ9 Causes Autosomal-Recessive Neonatal-Onset Primary Coenzyme Q10 Deficiency: A Potentially Treatable Form of Mitochondrial Disease. <i>American Journal of Human Genetics</i> , 2009, 84, 558-566.	6.2	206
34	Common Variation in the $\beta$ -Carotene 15,15 $\alpha$ -Monooxygenase 1 Gene Affects Circulating Levels of Carotenoids: A Genome-wide Association Study. <i>American Journal of Human Genetics</i> , 2009, 84, 123-133.	6.2	203
35	Genetic screening of Alzheimer's disease genes in Iberian and African samples yields novel mutations in presenilins and APP. <i>Neurobiology of Aging</i> , 2010, 31, 725-731.	3.1	196
36	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. <i>Lancet Neurology</i> , The, 2018, 17, 64-74.	10.2	195

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37	CSF biomarkers associated with disease heterogeneity in early Parkinson's disease: the Parkinson's Progression Markers Initiative study. <i>Acta Neuropathologica</i> , 2016, 131, 935-949.	7.7	190
38	Parkinson's disease and $\alpha$ -synuclein expression. <i>Movement Disorders</i> , 2011, 26, 2160-2168.	3.9	186
39	Genetic analysis implicates APOE, SNCA and suggests lysosomal dysfunction in the etiology of dementia with Lewy bodies. <i>Human Molecular Genetics</i> , 2014, 23, 6139-6146.	2.9	178
40	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. <i>Lancet Neurology</i> , 2007, 6, 414-420.	10.2	175
41	Whole Genome Analyses Suggest Ischemic Stroke and Heart Disease Share an Association With Polymorphisms on Chromosome 9p21. <i>Stroke</i> , 2008, 39, 1586-1589.	2.0	153
42	Estimating the causal influence of body mass index on risk of Parkinson disease: A Mendelian randomisation study. <i>PLoS Medicine</i> , 2017, 14, e1002314.	8.4	152
43	A genome-wide association analysis of serum iron concentrations. <i>Blood</i> , 2010, 115, 94-96.	1.4	142
44	Longitudinal Change of Clinical and Biological Measures in Early Parkinson's Disease: Parkinson's Progression Markers Initiative Cohort. <i>Movement Disorders</i> , 2018, 33, 771-782.	3.9	136
45	Spinocerebellar Ataxia Type 3 Phenotypically Resembling Parkinson Disease in a Black Family. <i>Archives of Neurology</i> , 2001, 58, 296.	4.5	135
46	Emerging pathways in genetic Parkinson's disease: Potential role of ceramide metabolism in Lewy body disease. <i>FEBS Journal</i> , 2008, 275, 5767-5773.	4.7	121
47	Multiple modality biomarker prediction of cognitive impairment in prospectively followed de novo Parkinson disease. <i>PLoS ONE</i> , 2017, 12, e0175674.	2.5	110
48	A Meta-analysis of Gene Expression Signatures of Blood Pressure and Hypertension. <i>PLoS Genetics</i> , 2015, 11, e1005035.	3.5	107
49	G2019S dardarin substitution is a common cause of Parkinson's disease in a Portuguese cohort. <i>Movement Disorders</i> , 2005, 20, 1653-1655.	3.9	106
50	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009, 18, 1524-1532.	2.9	106
51	The dardarin G2019S mutation is a common cause of Parkinson's disease but not other neurodegenerative diseases. <i>Neuroscience Letters</i> , 2005, 389, 137-139.	2.1	105
52	Tyrosinase exacerbates dopamine toxicity but is not genetically associated with Parkinson's disease. <i>Journal of Neurochemistry</i> , 2005, 93, 246-256.	3.9	103
53	The tau H2 haplotype is almost exclusively Caucasian in origin. <i>Neuroscience Letters</i> , 2004, 369, 183-185.	2.1	102
54	Genes and parkinsonism. <i>Lancet Neurology</i> , 2003, 2, 221-228.	10.2	98

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55	Complete screening for glucocerebrosidase mutations in Parkinson disease patients from Portugal. <i>Neurobiology of Aging</i> , 2009, 30, 1515-1517.	3.1	97
56	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. <i>Npj Parkinson's Disease</i> , 2019, 5, 8.	5.3	95
57	Clinical and dopamine transporter imaging characteristics of non-manifest LRRK2 and GBA mutation carriers in the Parkinson's Progression Markers Initiative (PPMI): a cross-sectional study. <i>Lancet Neurology</i> , The, 2020, 19, 71-80.	10.2	94
58	Phenomenology of ?Lubag? or X-linked dystonia-parkinsonism. <i>Movement Disorders</i> , 2002, 17, 1271-1277.	3.9	92
59	The law of mass action applied to neurodegenerative disease: a hypothesis concerning the etiology and pathogenesis of complex diseases. <i>Human Molecular Genetics</i> , 2004, 13, 123R-126.	2.9	86
60	Exome sequencing reveals an unexpected genetic cause of disease: NOTCH3 mutation in a Turkish family with Alzheimer's disease. <i>Neurobiology of Aging</i> , 2012, 33, 1008.e17-1008.e23.	3.1	86
61	Application of Genome-Wide Single Nucleotide Polymorphism Typing: Simple Association and Beyond. <i>PLoS Genetics</i> , 2006, 2, e150.	3.5	85
62	Human ataxias: a genetic dissection of inositol triphosphate receptor (ITPR1)-dependent signaling. <i>Trends in Neurosciences</i> , 2010, 33, 211-219.	8.6	81
63	The Evolution of Genetics: Alzheimer's and Parkinson's Diseases. <i>Neuron</i> , 2016, 90, 1154-1163.	8.1	81
64	Parkinson's disease and dementia with Lewy bodies: a difference in dose?. <i>Lancet</i> , The, 2004, 364, 1105-1107.	13.7	80
65	A generalizable hypothesis for the genetic architecture of disease: pleomorphic risk loci. <i>Human Molecular Genetics</i> , 2011, 20, R158-R162.	2.9	79
66	Genome-wide analysis of genetic correlation in dementia with Lewy bodies, Parkinson's and Alzheimer's diseases. <i>Neurobiology of Aging</i> , 2016, 38, 214.e7-214.e10.	3.1	78
67	Ethnic differences in the expression of neurodegenerative disease: Machado-Joseph disease in Africans and Caucasians. <i>Movement Disorders</i> , 2002, 17, 1068-1071.	3.9	77
68	Primary hyperhidrosis. <i>Clinical Autonomic Research</i> , 2003, 13, 96-98.	2.5	74
69	Torsin A haplotype predisposes to idiopathic dystonia. <i>Annals of Neurology</i> , 2005, 57, 765-767.	5.3	73
70	Analysis of an early-onset Parkinson's disease cohort for DJ-1 mutations. <i>Movement Disorders</i> , 2004, 19, 796-800.	3.9	71
71	Analysis of IFT74 as a candidate gene for chromosome 9p-linked ALS-FTD. <i>BMC Neurology</i> , 2006, 6, 44.	1.8	70
72	Linkage, whole genome sequence, and biological data implicate variants in RAB10 in Alzheimer's disease resilience. <i>Genome Medicine</i> , 2017, 9, 100.	8.2	67

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73	SCA15 Due to Large ITPR1 Deletions in a Cohort of 333 White Families With Dominant Ataxia. Archives of Neurology, 2011, 68, 637-43.	4.5	65
74	Glucocerebrosidase mutations are also found in subjects with early-onset parkinsonism from Venezuela. Movement Disorders, 2006, 21, 282-283.	3.9	64
75	Familial Parkinsonism and early onset Parkinson's disease in a Brazilian movement disorders clinic: Phenotypic characterization and frequency of <i>SNCA</i>, <i>PRKN</i>, <i>PINK1</i>, and <i>LRRK2</i> mutations. Movement Disorders, 2009, 24, 662-666.	3.9	63
76	How genetics research in Parkinson's disease is enhancing understanding of the common idiopathic forms of the disease. Current Opinion in Neurology, 2005, 18, 706-711.	3.6	62
77	Extracting Vertical Displacement Rates in Shanghai (China) with Multi-Platform SAR Images. Remote Sensing, 2015, 7, 9542-9562.	4.0	62
78	Analysis of Nigerians with Apparently Sporadic Parkinson Disease for Mutations in LRRK2, PRKN and ATXN3. PLoS ONE, 2008, 3, e3421.	2.5	61
79	TDP-43 Is Not a Common Cause of Sporadic Amyotrophic Lateral Sclerosis. PLoS ONE, 2008, 3, e2450.	2.5	60
80	A systematic screening to identify<i>de novo</i> mutations causing sporadic early-onset Parkinson's disease. Human Molecular Genetics, 2015, 24, 6711-6720.	2.9	59
81	A Rare Truncating Mutation in ADH1C (G78Stop) Shows Significant Association With Parkinson Disease in a Large International Sample. Archives of Neurology, 2005, 62, 74.	4.5	57
82	Next generation sequencing techniques in neurological diseases: redefining clinical and molecular associations. Human Molecular Genetics, 2014, 23, R47-R53.	2.9	57
83	Advancing age is associated with gene expression changes resembling mTOR inhibition: Evidence from two human populations. Mechanisms of Ageing and Development, 2012, 133, 556-562.	4.6	54
84	Investigating the role of rare coding variability in Mendelian dementia genes ( APP , PSEN1 , PSEN2 , GRN) Tj ETQq0,0,0 rgBT /Qverlock 1	3.1	53
85	Analysis of Parkinson disease patients from Portugal for mutations in SNCA, PRKN, PINK1 and LRRK2. BMC Neurology, 2008, 8, 1.	1.8	52
86	Genetic Variability in CLU and Its Association with Alzheimer's Disease. PLoS ONE, 2010, 5, e9510.	2.5	52
87	Conflicting Results Regarding the Semaphorin Gene (SEMA5A) and the Risk for Parkinson Disease. American Journal of Human Genetics, 2006, 78, 1082-1083.	6.2	50
88	Sequential Use of Transcriptional Profiling, Expression Quantitative Trait Mapping, and Gene Association Implicates MMP20 in Human Kidney Aging. PLoS Genetics, 2009, 5, e1000685.	3.5	50
89	Association of Î±-synuclein Rep1 polymorphism and Parkinson's disease: Influence of Rep1 on age at onset. Movement Disorders, 2006, 21, 534-539.	3.9	49
90	Genome-wide association studies in neurological disorders. Lancet Neurology, The, 2008, 7, 1067-1072.	10.2	49

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91	A genome-wide screening and SNPs-to-genes approach to identify novel genetic risk factors associated with frontotemporal dementia. <i>Neurobiology of Aging</i> , 2015, 36, 2904.e13-2904.e26.	3.1	48
92	Shared biological pathways for frailty and cognitive impairment: A systematic review. <i>Ageing Research Reviews</i> , 2018, 47, 149-158.	10.9	48
93	Clinical and Dopamine Transporter Imaging Characteristics of Leucine Rich Repeat Kinase 2 (LRRK2) and Glucosylceramidase Beta (GBA) Parkinson's Disease Participants in the Parkinson's Progression Markers Initiative: A Cross-sectional Study. <i>Movement Disorders</i> , 2020, 35, 833-844.	3.9	48
94	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019, 34, 1851-1863.	3.9	47
95	Refinement of the PARK3 locus on chromosome 2p13 and the analysis of 14 candidate genes. <i>European Journal of Human Genetics</i> , 2001, 9, 659-666.	2.8	46
96	Genomewide SNP assay reveals mutations underlying Parkinson disease. <i>Human Mutation</i> , 2008, 29, 315-322.	2.5	46
97	Candidate Gene Polymorphisms for Ischemic Stroke. <i>Stroke</i> , 2009, 40, 3436-3442.	2.0	46
98	Genetics of early-onset Parkinson's disease in Finland: exome sequencing and genome-wide association study. <i>Neurobiology of Aging</i> , 2017, 53, 195.e7-195.e10.	3.1	46
99	Identification of PSEN1 and PSEN2 gene mutations and variants in Turkish dementia patients. <i>Neurobiology of Aging</i> , 2012, 33, 1850.e17-1850.e27.	3.1	44
100	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. <i>Annals of Human Genetics</i> , 2013, 77, 85-105.	0.8	41
101	Identical twins with the <i>C9orf72</i> repeat expansion are discordant for ALS. <i>Neurology</i> , 2014, 83, 1476-1478.	1.1	40
102	Characterization of Recessive Parkinson Disease in a Large Multicenter Study. <i>Annals of Neurology</i> , 2020, 88, 843-850.	5.3	40
103	A <i>C6orf10/LOC101929163</i> locus is associated with age of onset in <i>C9orf72</i> carriers. <i>Brain</i> , 2018, 141, 2895-2907.	7.6	39
104	An evaluation of the Team Objective Structured Clinical Examination (TOSCE). <i>Medical Education</i> , 1999, 33, 34-41.	2.1	37
105	X-linked dystonia (aka Lubag) presenting predominantly with parkinsonism: A more benign phenotype?. <i>Movement Disorders</i> , 2002, 17, 200-202.	3.9	37
106	Genetic susceptibility in Parkinson's disease. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2009, 1792, 597-603.	3.8	37
107	Mutation analysis of patients with neurodegenerative disorders using NeuroX array. <i>Neurobiology of Aging</i> , 2015, 36, 545.e9-545.e14.	3.1	36
108	The <i>TOR1A</i> polymorphism rs1182 and the risk of spread in primary blepharospasm. <i>Movement Disorders</i> , 2009, 24, 613-616.	3.9	35

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109	Genome-wide assessment of Parkinson's disease in a Southern Spanish population. <i>Neurobiology of Aging</i> , 2016, 45, 213.e3-213.e9.	3.1	35
110	Anticholinergic Drug Induced Cognitive and Physical Impairment: Results from the InCHIANTI Study. <i>Journals of Gerontology - Series A Biological Sciences and Medical Sciences</i> , 2020, 75, 995-1002.	3.6	34
111	Anatomy of Subsidence in Tianjin from Time Series InSAR. <i>Remote Sensing</i> , 2016, 8, 266.	4.0	33
112	Parkin disease in a Brazilian kindred: Manifesting heterozygotes and clinical follow-up over 10 years. <i>Movement Disorders</i> , 2005, 20, 479-484.	3.9	32
113	Clinical features, with video documentation, of the original familial lewy body parkinsonism caused by Î±-synuclein triplication (Iowa kindred). <i>Movement Disorders</i> , 2011, 26, 2134-2136.	3.9	32
114	Novel progranulin mutation: Screening for PGRN mutations in a Portuguese series of FTD/CBS cases. <i>Movement Disorders</i> , 2008, 23, 1269-1273.	3.9	30
115	Polygenic risk score in postmortem diagnosed sporadic early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2018, 62, 244.e1-244.e8.	3.1	30
116	NOTCH3 Variants and Risk of Ischemic Stroke. <i>PLoS ONE</i> , 2013, 8, e75035.	2.5	30
117	Heritability and genetic variance of dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2019, 127, 492-501.	4.4	29
118	Exome sequencing identifies 2 novel presenilin 1 mutations (p.L166V and p.S230R) in British early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014, 35, 2422.e13-2422.e16.	3.1	28
119	Shared mechanisms for cognitive impairment and physical frailty: A model for complex systems. <i>Alzheimer's and Dementia: Translational Research and Clinical Interventions</i> , 2020, 6, e12027.	3.7	28
120	Genetic and Phenotypic Basis of Autosomal Dominant Parkinson's Disease in a Large Multi-Center Cohort. <i>Frontiers in Neurology</i> , 2020, 11, 682.	2.4	28
121	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. <i>Acta Neuropathologica Communications</i> , 2020, 8, 5.	5.2	27
122	Paraoxonase 1 (PON1) gene polymorphisms and Parkinson's disease in a Finnish population. <i>Neuroscience Letters</i> , 2004, 367, 168-170.	2.1	26
123	Whole blood gene expression and interleukin-6 levels. <i>Genomics</i> , 2014, 104, 490-495.	2.9	24
124	Linkage Disequilibrium and Association Analysis of Î²-Synuclein and Alcohol and Drug Dependence in Two American Indian Populations. <i>Alcoholism: Clinical and Experimental Research</i> , 2007, 31, 070212174136004-???	2.4	23
125	A thorough assessment of benign genetic variability in GRN and MAPT. <i>Human Mutation</i> , 2010, 31, E1126-E1140.	2.5	23
126	Gene expression markers of age-related inflammation in two human cohorts. <i>Experimental Gerontology</i> , 2015, 70, 37-45.	2.8	23



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127	Longitudinal Measurements of Glucocerebrosidase activity in Parkinson's patients. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1816-1830.	3.7	23
128	Neurofibrillary tau pathology modulated by genetic variation of <i>SNCA</i> . <i>Annals of Neurology</i> , 2008, 64, 348-352.	5.3	22
129	Kinase signaling pathways as potential targets in the treatment of Parkinson's disease. <i>Expert Review of Proteomics</i> , 2007, 4, 783-792.	3.0	21
130	Mutation analysis of sporadic early-onset Alzheimer's disease using the NeuroX array. <i>Neurobiology of Aging</i> , 2017, 49, 215.e1-215.e8.	3.1	21
131	Ethnic Differences and Disease Phenotypes. <i>Science</i> , 2003, 300, 739-740.	12.6	20
132	A consanguineous Turkish family with early-onset Parkinson's disease and an exon 4 parkin deletion. <i>Movement Disorders</i> , 2004, 19, 812-816.	3.9	20
133	Multiple system atrophy: the application of genetics in understanding etiology. <i>Clinical Autonomic Research</i> , 2015, 25, 19-36.	2.5	20
134	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. <i>PLoS ONE</i> , 2016, 11, e0162592.	2.5	19
135	Smell testing is abnormal in <i>LUBAG</i> or X-linked dystonia-parkinsonism: a pilot study. <i>Parkinsonism and Related Disorders</i> , 2004, 10, 407-410.	2.2	18
136	Defining the ends of Parkin exon 4 deletions in two different families with Parkinson's disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2005, 133B, 120-123.	1.7	18
137	Mutation at the SCA17 locus is not a common cause of parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2003, 9, 317-320.	2.2	15
138	APOE and A $\beta$ PP Gene Variation in Cortical and Cerebrovascular Amyloid- $\beta$ Pathology and Alzheimer's Disease: A Population-Based Analysis. <i>Journal of Alzheimer's Disease</i> , 2011, 26, 377-385.	2.6	15
139	Feasibility and safety of lumbar puncture in the Parkinson's disease research participants: Parkinson's Progression Marker Initiative (PPMI). <i>Parkinsonism and Related Disorders</i> , 2019, 62, 201-209.	2.2	15
140	Genomic Risk Profiling of Ischemic Stroke: Results of an International Genome-Wide Association Meta-Analysis. <i>PLoS ONE</i> , 2011, 6, e23161.	2.5	14
141	Blepharospasm: A genetic screening study in 132 patients. <i>Parkinsonism and Related Disorders</i> , 2019, 64, 315-318.	2.2	13
142	A comprehensive screening of copy number variability in dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2019, 75, 223.e1-223.e10.	3.1	13
143	Amotrophic Lateral Sclerosis: An Emerging Era of Collaborative Gene Discovery. <i>PLoS ONE</i> , 2007, 2, e1254.	2.5	13
144	Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. <i>Neurobiology of Aging</i> , 2017, 49, 214.e13-214.e15.	3.1	12

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145	The Future of Genetic Analysis of Neurological Disorders. <i>Neurobiology of Disease</i> , 2000, 7, 65-69.	4.4	11
146	Peripheral GRN mRNA and Serum Progranulin Levels as a Potential Indicator for Both the Presence of Splice Site Mutations and Individuals at Risk for Frontotemporal Dementia. <i>Journal of Alzheimer's Disease</i> , 2019, 67, 159-167.	2.6	11
147	Clinical Variability of SYNJ1-Associated Early-Onset Parkinsonism. <i>Frontiers in Neurology</i> , 2021, 12, 648457.	2.4	11
148	A Bayesian mathematical model of motor and cognitive outcomes in Parkinson's disease. <i>PLoS ONE</i> , 2017, 12, e0178982.	2.5	11
149	Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , 2015, 24, 1504-1512.	2.9	8
150	No evidence for tau duplications in frontal temporal dementia families showing genetic linkage to the tau locus in which tau mutations have not been found. <i>Neuroscience Letters</i> , 2004, 363, 99-101.	2.1	7
151	The HapMap. <i>Archives of Neurology</i> , 2008, 65, 319-21.	4.5	7
152	Splicing factor 3B1 hypomethylation is associated with altered SF3B1 transcript expression in older humans. <i>Mechanisms of Ageing and Development</i> , 2014, 135, 50-56.	4.6	7
153	Exploring dementia and neuronal ceroid lipofuscinosis genes in 100 FTD-like patients from 6 towns and rural villages on the Adriatic Sea coast of Apulia. <i>Scientific Reports</i> , 2021, 11, 6353.	3.3	7
154	Whole genome association studies: Deciding when persistence becomes perseverance. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2008, 147B, 131-133.	1.7	6
155	Using Exome Sequencing to Reveal Mutations in TREM2 Presenting as a Frontotemporal Dementia-like Syndrome Without Bone Involvement. <i>JAMA Neurology</i> , 0, , 1.	9.0	6
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161	Prion genotypes in Central America suggest selection for the V129 allele. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2006, 141B, 33-35.	1.7	3
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163	Familiality in simple and complex disease. <i>Clinical Autonomic Research</i> , 2003, 13, 88-90.	2.5	1
164	A simple and efficient algorithm for genome-wide homozygosity analysis in disease. <i>Molecular Systems Biology</i> , 2009, 5, 304.	7.2	1