Jeffery M Vance

List of Publications by Year
in descending order

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241 papers	23,480 citations	12330 69 h-index	8630 146 g-index
267 all docs	267 docs citations	267 times ranked	28747 citing authors

#	Article	IF	Citations
1	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	21.4	1,962
2	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. Nature Genetics, 2004, 36, 449-451.	21.4	1,391
3	The Deacetylase HDAC6 Regulates Aggresome Formation and Cell Viability in Response to Misfolded Protein Stress. Cell, 2003, 115, 727-738.	28.9	1,349
4	A Mutation in the TRPC6 Cation Channel Causes Familial Focal Segmental Glomerulosclerosis. Science, 2005, 308, 1801-1804.	12.6	967
5	New insights into the genetic etiology of Alzheimerâ€™s disease and related dementias. Nature Genetics, 2022, 54, 412-436.	21.4	700
6	PGC-1 Î±, A Potential Therapeutic Target for Early Intervention in Parkinsonâ€™s Disease. Science Translational Medicine, 2010, 2, 52ra73.	12.4	691
7	Mitochondrial Polymorphisms Significantly Reduce the Risk of Parkinson Disease. American Journal of Human Genetics, 2003, 72, 804-811.	6.2	507
8	Glycyl tRNA Synthetase Mutations in Charcot-Marie-Tooth Disease Type 2D and Distal Spinal Muscular Atrophy Type V. American Journal of Human Genetics, 2003, 72, 1293-1299.	6.2	505
9	Comprehensive Research Synopsis and Systematic Meta-Analyses in Parkinson's Disease Genetics: The PDGene Database. PLoS Genetics, 2012, 8, e1002548.	3.5	495
10	Molecular markers of early Parkinson's disease based on gene expression in blood. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 955-960.	7.1	462
11	No Gene Is an Island: The Flip-Flop Phenomenon. American Journal of Human Genetics, 2007, 80, 531-538.	6.2	437
12	Variation in the miRNA-433 Binding Site of FGF20 Confers Risk for Parkinson Disease by Overexpression of Î±-Synuclein. American Journal of Human Genetics, 2008, 82, 283-289.	6.2	437
13	Huntingtin and DRPLA proteins selectively interact with the enzyme GAPDH. Nature Medicine, 1996, 2, 347-350.	30.7	429
14	Genomeâ€Wide Association Study Confirms SNPs in SNCA and the MAPT Region as Common Risk Factors for Parkinson Disease. Annals of Human Genetics, 2010, 74, 97-109.	0.8	417
15	Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. Annals of Neurology, 2006, 59, 276-281.	5.3	380
16	A conserved sorting-associated protein is mutant in chorea-acanthocytosis. Nature Genetics, 2001, 28, 119-120.	21.4	357
17	MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. Brain, 2006, 129, 2093-2102.	7.6	351
18	Ganglioside-induced differentiation-associated protein-1 is mutant in Charcot-Marie-Tooth disease type 4A/8q21. Nature Genetics, 2002, 30, 21-22.	21.4	348

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19	SNPing Away at Complex Diseases: Analysis of Single-Nucleotide Polymorphisms around APOE in Alzheimer Disease. American Journal of Human Genetics, 2000, 67, 383-394.	6.2	342
20	Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. Nature Genetics, 2005, 37, 289-294.	21.4	324
21	Age at Onset in Two Common Neurodegenerative Diseases Is Genetically Controlled. American Journal of Human Genetics, 2002, 70, 985-993.	6.2	291
22	Functional variants in the LRRK2 gene confer shared effects on risk for Crohnâ€™s disease and Parkinsonâ€™s disease. Science Translational Medicine, 2018, 10, .	12.4	273
23	Analysis of European mitochondrial haplogroups with Alzheimer disease risk. Neuroscience Letters, 2004, 365, 28-32.	2.1	264
24	Metaâ€analysis of Parkinson's Disease: Identification of a novel locus, RIT2. Annals of Neurology, 2012, 71, 370-384.	5.3	264
25	Complete Genomic Screen in Parkinson Disease. JAMA - Journal of the American Medical Association, 2001, 286, 2239.	7.4	257
26	Convergence of miRNA Expression Profiling, Î±-Synuclein Interacton and GWAS in Parkinson's Disease. PLoS ONE, 2011, 6, e25443.	2.5	235
27	The Haw River Syndrome: Dentatorubropallidoluysian atrophy (DRPLA) in an Africanâ€“American family. Nature Genetics, 1994, 7, 521-524.	21.4	228
28	Genomic screen and follow-up analysis for autistic disorder. American Journal of Medical Genetics Part A, 2002, 114, 99-105.	2.4	226
29	Pesticide exposure and risk of Parkinson's disease: A family-based case-control study. BMC Neurology, 2008, 8, 6.	1.8	221
30	Identification of MeCP2 mutations in a series of females with autistic disorder. Pediatric Neurology, 2003, 28, 205-211.	2.1	210
31	Mutations in the Novel Mitochondrial Protein REEP1 Cause Hereditary Spastic Paraplegia Type 31. American Journal of Human Genetics, 2006, 79, 365-369.	6.2	209
32	Glutathione S-transferase omega-1 modifiesage-at-onset of Alzheimer disease and Parkinson disease. Human Molecular Genetics, 2003, 12, 3259-3267.	2.9	208
33	Genome-Wide Gene-Environment Study Identifies Glutamate Receptor Gene GRIN2A as a Parkinson's Disease Modifier Gene via Interaction with Coffee. PLoS Genetics, 2011, 7, e1002237.	3.5	206
34	Parkin mutations and susceptibility alleles in lateâ€onset Parkinson's disease. Annals of Neurology, 2003, 53, 624-629.	5.3	201
35	A noise-reduction GWAS analysis implicates altered regulation of neurite outgrowth and guidance in autism. Molecular Autism, 2011, 2, 1.	4.9	191
36	Linkage of Tunisian autosomal recessive Duchenneâ€“like muscular dystrophy to the pericentromeric region of chromosome 13q. Nature Genetics, 1992, 2, 315-317.	21.4	186

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37	Localization of a Gene (CMT2A) for Autosomal Dominant Charcot-Marie-Tooth Disease Type 2 to Chromosome 1p and Evidence of Genetic Heterogeneity. Genomics, 1993, 17, 370-375.	2.9	173
38	North Carolina macular dystrophy is assigned to chromosome 6. Genomics, 1992, 13, 681-685.	2.9	167
39	Whole-Exome Sequencing Links a Variant in DHDDS to Retinitis Pigmentosa. American Journal of Human Genetics, 2011, 88, 201-206.	6.2	155
40	A Genomewide Scan for Early-Onset Coronary Artery Disease in 438 Families: The GENECARD Study. American Journal of Human Genetics, 2004, 75, 436-447.	6.2	152
41	Mutations in the ER-shaping protein reticulon 2 cause the axon-degenerative disorder hereditary spastic paraplegia type 12. Journal of Clinical Investigation, 2012, 122, 538-544.	8.2	149
42	Expression Profiling of Substantia Nigra in Parkinson Disease, Progressive Supranuclear Palsy, and Frontotemporal Dementia With Parkinsonism. Archives of Neurology, 2005, 62, 917-21.	4.5	146
43	Identification of TMEM230 mutations in familial Parkinson's disease. Nature Genetics, 2016, 48, 733-739.	21.4	146
44	Fibroblast Growth Factor 20 Polymorphisms and Haplotypes Strongly Influence Risk of Parkinson Disease. American Journal of Human Genetics, 2004, 74, 1121-1127.	6.2	136
45	Identification and Expression Analysis of Spastin Gene Mutations in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2001, 68, 1077-1085.	6.2	130
46	Dementia Revealed: Novel Chromosome 6 Locus for Late-Onset Alzheimer Disease Provides Genetic Evidence for Folate-Pathway Abnormalities. PLoS Genetics, 2010, 6, e1001130.	3.5	130
47	Association of Polymorphisms in the Apolipoprotein E Region with Susceptibility to and Progression of Multiple Sclerosis. American Journal of Human Genetics, 2002, 70, 708-717.	6.2	125
48	Linkage of a Gene Causing Familial Focal Segmental Glomerulosclerosis to Chromosome 11 and Further Evidence of Genetic Heterogeneity. Genomics, 1999, 58, 113-120.	2.9	117
49	Ancestral origin of ApoE Îµ4 Alzheimer disease risk in Puerto Rican and African American populations. PLoS Genetics, 2018, 14, e1007791.	3.5	117
50	Peakwide Mapping on Chromosome 3q13 Identifies the Kalirin Gene as a Novel Candidate Gene for Coronary Artery Disease. American Journal of Human Genetics, 2007, 80, 650-663.	6.2	110
51	Smoking, Caffeine, and Nonsteroidal Anti-inflammatory Drugs in Families With Parkinson Disease. Archives of Neurology, 2007, 64, 576.	4.5	107
52	Exome sequencing allows for rapid gene identification in a Charcotâ€Marieâ€Tooth family. Annals of Neurology, 2011, 69, 464-470.	5.3	107
53	A Loss-of-Function Variant in the Human Histidyl-tRNA Synthetase (HARS) Gene is Neurotoxic In Vivo. Human Mutation, 2013, 34, 191-199.	2.5	104
54	SNPselector: a web tool for selecting SNPs for genetic association studies. Bioinformatics, 2005, 21, 4181-4186.	4.1	101

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55	Ordered-Subsets Linkage Analysis Detects Novel Alzheimer Disease Loci on Chromosomes 2q34 and 15q22. American Journal of Human Genetics, 2003, 73, 1041-1051.	6.2	99
56	SRRM2, a Potential Blood Biomarker Revealing High Alternative Splicing in Parkinson's Disease. PLoS ONE, 2010, 5, e9104.	2.5	97
57	Vitamin D Receptor Gene as a Candidate Gene for Parkinson Disease. Annals of Human Genetics, 2011, 75, 201-210.	0.8	95
58	myotilin Mutation Found in Second Pedigree with LGMD1A. American Journal of Human Genetics, 2002, 71, 1428-1432.	6.2	92
59	Nitric oxide synthase genes and their interactions with environmental factors in Parkinsonâ€™s disease. Neurogenetics, 2008, 9, 249-262.	1.4	91
60	Notch activation induces endothelial cell senescence and pro-inflammatory response: Implication of Notch signaling in atherosclerosis. Atherosclerosis, 2012, 225, 296-303.	0.8	90
61	A Recurrent loss-of-function alanyl-tRNA synthetase (AARSâ€‰) mutation in patients with charcot-marie-tooth disease type 2N (CMT2N). Human Mutation, 2012, 33, 244-253.	2.5	90
62	Mechanisms of Disease: a molecular genetic update on hereditary axonal neuropathies. Nature Clinical Practice Neurology, 2006, 2, 45-53.	2.5	88
63	Three probands with autistic disorder and isodicentric chromosome 15. American Journal of Medical Genetics Part A, 2000, 96, 365-372.	2.4	87
64	Neuropeptide Y Gene Polymorphisms Confer Risk of Early-Onset Atherosclerosis. PLoS Genetics, 2009, 5, e1000318.	3.5	87
65	The mtDNA Mutation Spectrum of the Progeroid Polg Mutator Mouse Includes Abundant Control Region Multimers. Cell Metabolism, 2010, 12, 675-682.	16.2	86
66	Identification of a New Autosomal Dominant Limb-Girdle Muscular Dystrophy Locus on Chromosome 7. American Journal of Human Genetics, 1999, 64, 556-562.	6.2	82
67	GATA2 Is Associated with Familial Early-Onset Coronary Artery Disease. PLoS Genetics, 2006, 2, e139.	3.5	82
68	Gene Expression Profiles in Parkinson Disease Prefrontal Cortex Implicate FOXO1 and Genes under Its Transcriptional Regulation. PLoS Genetics, 2012, 8, e1002794.	3.5	76
69	ABCA7 frameshift deletion associated with Alzheimer disease in African Americans. Neurology: Genetics, 2016, 2, e79.	1.9	74
70	Identifying Consensus Disease Pathways in Parkinson's Disease Using an Integrative Systems Biology Approach. PLoS ONE, 2011, 6, e16917.	2.5	72
71	Localization of Charcot-Marie-Tooth disease type 1a (CMT1A) to chromosome 17p11.2. Genomics, 1991, 9, 623-628.	2.9	70
72	Genetic Complexity and Parkinson's Disease. Science, 1997, 277, 387-390.	12.6	70

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73	Identification of a New Locus for Autosomal Recessive Charcotâ€“Marieâ€“Tooth Disease with Focally Folded Myelin on Chromosome 11p15. Genomics, 1999, 62, 344-349.	2.9	69
74	C9ORF72 Intermediate Repeat Copies Are a Significant Risk Factor for Parkinson Disease. Annals of Human Genetics, 2013, 77, 351-363.	0.8	69
75	Whole exome sequencing of rare variants in EIF4G1 and VPS35 in Parkinson disease. Neurology, 2013, 80, 982-989.	1.1	68
76	Association between the neuron-specific RNA-binding protein ELAVL4 and Parkinson disease. Human Genetics, 2005, 117, 27-33.	3.8	67
77	Identification of Risk and Age-at-Onset Genes on Chromosome 1p in Parkinson Disease. American Journal of Human Genetics, 2005, 77, 252-264.	6.2	67
78	Convergent Pathways in Idiopathic Autism Revealed by Time Course Transcriptomic Analysis of Patient-Derived Neurons. Scientific Reports, 2018, 8, 8423.	3.3	67
79	Molecular genetics of autosomal-dominant axonal Charcot-Marie-Tooth disease. NeuroMolecular Medicine, 2006, 8, 63-74.	3.4	66
80	Comprehensive genetic analysis of the platelet activating factor acetylhydrolase (PLA2G7) gene and cardiovascular disease in caseâ€“control and family datasets. Human Molecular Genetics, 2008, 17, 1318-1328.	2.9	66
81	Identification of genetic polymorphisms associated with risk for pulmonary hypertension in sickle cell disease. Blood, 2008, 111, 5721-5726.	1.4	66
82	Comparison of Three Targeted Enrichment Strategies on the SOLiD Sequencing Platform. PLoS ONE, 2011, 6, e18595.	2.5	66
83	Genetic polymorphisms associated with priapism in sickle cell disease. British Journal of Haematology, 2007, 137, 262-267.	2.5	64
84	Derivation of autism spectrum disorder-specific induced pluripotent stem cells from peripheral blood mononuclear cells. Neuroscience Letters, 2012, 516, 9-14.	2.1	64
85	Geneâ€“environment interactions in Parkinson's disease and other forms of parkinsonism. NeuroToxicology, 2010, 31, 598-602.	3.0	63
86	Vitamin D from different sources is inversely associated with Parkinson disease. Movement Disorders, 2015, 30, 560-566.	3.9	61
87	Analysis of Association at Single Nucleotide Polymorphisms in the APOE Region. Genomics, 2000, 63, 7-12.	2.9	60
88	Revealing the role of glutathione S-transferase omega in age-at-onset of Alzheimer and Parkinson diseases. Neurobiology of Aging, 2006, 27, 1087-1093.	3.1	60
89	The Many Faces of Charcot-Marie-Tooth Disease. Archives of Neurology, 2000, 57, 638.	4.5	54
90	Association Study of Parkin Gene Polymorphisms With Idiopathic Parkinson Disease. Archives of Neurology, 2003, 60, 975.	4.5	51

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91	Genetic effects in the leukotriene biosynthesis pathway and association with atherosclerosis. Human Genetics, 2009, 125, 217-229.	3.8	51
92	Genome-wide brain DNA methylation analysis suggests epigenetic reprogramming in Parkinson disease. Neurology: Genetics, 2019, 5, e342.	1.9	50
93	A Duplication in Chromosome 4q35 Is Associated with Hereditary Benign Intraepithelial Dyskeratosis. American Journal of Human Genetics, 2001, 68, 491-494.	6.2	49
94	Genomic convergence to identify candidate genes for Parkinson disease: SAGE analysis of the substantia nigra. Movement Disorders, 2005, 20, 1299-1309.	3.9	48
95	PARK10 is a major locus for sporadic neuropathologically confirmed Parkinson disease. Neurology, 2015, 84, 972-980.	1.1	48
96	The transcription factor orthodenticle homeobox 2 influences axonal projections and vulnerability of midbrain dopaminergic neurons. Brain, 2010, 133, 2022-2031.	7.6	47
97	Thrombotic Storm Revisited: Preliminary Diagnostic Criteria Suggested by the Thrombotic Storm Study Group. American Journal of Medicine, 2011, 124, 290-296.	1.5	45
98	Toxicity of expanded polyglutamine-domain proteins in Escherichia coli. FEBS Letters, 1996, 399, 135-139.	2.8	44
99	Clinical and genetic heterogeneity in familial focal segmental glomerulosclerosis. Kidney International, 1999, 55, 1241-1246.	5.2	44
100	Genomic convergence: identifying candidate genes for Parkinson's disease by combining serial analysis of gene expression and genetic linkage. Human Molecular Genetics, 2003, 12, 671-7.	2.9	44
101	A genome-wide linkage analysis of dementia in the Amish. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 160-166.	1.7	42
102	Segregation of a rare TTC3 variant in an extended family with late-onset Alzheimer disease. Neurology: Genetics, 2016, 2, e41.	1.9	41
103	Early-Onset Alzheimer Disease and Candidate Risk Genes Involved in Endolysosomal Transport. JAMA Neurology, 2017, 74, 1113.	9.0	41
104	A genome-wide search for linkage to asthma phenotypes in the genetics of asthma international network families: evidence for a major susceptibility locus on chromosome 2p. European Journal of Human Genetics, 2006, 14, 307-316.	2.8	40
105	NOS2Aand the modulating effect of cigarette smoking in Parkinson's disease. Annals of Neurology, 2006, 60, 366-373.	5.3	38
106	Confirmation of linkage of hereditary partial lipodystrophy to chromosome 1q21-22. , 1999, 82, 161-165.		33
107	Family-based caseâ€“control study of MAOA and MAOB polymorphisms in Parkinson disease. Movement Disorders, 2006, 21, 2175-2180.	3.9	33
108	Increased APOE Îµ4 expression is associated with the difference in Alzheimer's disease risk from diverse ancestral backgrounds. Alzheimer's and Dementia, 2021, 17, 1179-1188.	0.8	33

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109	Chorea-acanthocytosis: A report of three new families and implications for genetic counselling. American Journal of Medical Genetics Part A, 1987, 28, 403-410.	2.4	32
110	Linkage of a Gene Causing Familial Membranoproliferative Glomerulonephritis Type III to Chromosome 1. Journal of the American Society of Nephrology: JASN, 2002, 13, 2052-2057.	6.1	32
111	Mutation screening of mitofusin 2 in Charcot-Marie-Tooth disease type 2. Journal of Neurology, 2011, 258, 1234-1239.	3.6	32
112	Overlap between Parkinson disease and Alzheimer disease in ABCA7 functional variants. Neurology: Genetics, 2016, 2, e44.	1.9	31
113	Linkage disequilibrium and haplotype tagging polymorphisms in the Tau H1 haplotype. Neurogenetics, 2004, 5, 147-155.	1.4	30
114	Genomewide Association Studies of <scp>LRRK2</scp> Modifiers of Parkinson's Disease. Annals of Neurology, 2021, 90, 76-88.	5.3	30
115	Emerging pathways for hereditary axonopathies. Journal of Molecular Medicine, 2005, 83, 935-943.	3.9	29
116	A rare novel deletion of the tyrosine hydroxylase gene in Parkinson disease. Human Mutation, 2010, 31, E1767-E1771.	2.5	29
117	Clinical causes and treatment of the thrombotic storm. Expert Review of Hematology, 2012, 5, 653-659.	2.2	29
118	Confirmation of linkage in von Hippel-Lindau disease. Genomics, 1990, 6, 565-567.	2.9	27
119	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. Journal of Neurology, 2010, 257, 735-741.	3.6	24
120	Absence of C9ORF72 expanded or intermediate repeats in autopsyâ€confirmed Parkinson's disease. Movement Disorders, 2014, 29, 827-830.	3.9	24
121	Autistic Disorder and Chromosome 15q11â€“q13: Construction and Analysis of a BAC/PAC Contig. Genomics, 1999, 62, 325-331.	2.9	23
122	Heterogeneity in Paget disease of the bone. American Journal of Medical Genetics Part A, 2000, 92, 303-307.	2.4	22
123	Comprehensive association analysis of APOE regulatory region polymorphisms in Alzheimer disease. Neurogenetics, 2004, 5, 201-208.	1.4	22
124	hVGAT-mCherry: A novel molecular tool for analysis of GABAergic neurons derived from human pluripotent stem cells. Molecular and Cellular Neurosciences, 2015, 68, 244-257.	2.2	22
125	Genetic Variants of Microtubule Actin Cross-linking Factor 1 (MACF1) Confer Risk for Parkinsonâ€™s Disease. Molecular Neurobiology, 2017, 54, 2878-2888.	4.0	22
126	Recombinant DNA strategies in genetic neurological diseases. Muscle and Nerve, 1983, 6, 339-355.	2.2	21

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127	Tyrosine Hydroxylase Gene: Another Piece of the Genetic Puzzle of Parkinsonâ€™s Disease. CNS and Neurological Disorders - Drug Targets, 2012, 11, 469-481.	1.4	21
128	RNA editing alterations in a multi-ethnic Alzheimer disease cohort converge on immune and endocytic molecular pathways. Human Molecular Genetics, 2019, 28, 3053-3061.	2.9	19
129	A locus at 19q13.31 significantly reduces the ApoE Îµ4 risk for Alzheimerâ€™s Disease in African Ancestry. PLoS Genetics, 2022, 18, e1009977.	3.5	19
130	Use of a CEPH Meiotic Breakpoint Panel to Refine the Locus of Limb-Girdle Muscular Dystrophy Type 1A (LGMD1A) to a 2-Mb Interval on 5q31. Genomics, 1998, 54, 250-255.	2.9	18
131	Different Mutations in Carbohydrate Sulfotransferase 6 (CHST6) Gene Cause Macular Corneal Dystrophy Types I and II in a Single Sibship. American Journal of Ophthalmology, 2005, 139, 1118-1120.	3.3	18
132	Response to Zaykin and Shibata. American Journal of Human Genetics, 2008, 82, 796-797.	6.2	18
133	Findings from a community education needs assessment to facilitate the integration of genomic medicine into primary care. Genetics in Medicine, 2010, 12, 587-593.	2.4	18
134	Identification of seven novel SNPS (five nucleotide and two amino acid substitutions) in the connexin31 (GJB3) gene. Human Mutation, 2000, 15, 481-482.	2.5	17
135	Detecting Genetic Interactions in Pathwayâ€Based Genomeâ€Wide Association Studies. Genetic Epidemiology, 2014, 38, 300-309.	1.3	17
136	Mutation K42E in Dehydrodolichol Diphosphate Synthase (DHDDS) Causes Recessive Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2014, 801, 165-170.	1.6	17
137	Genomic convergence: identifying candidate genes for Parkinson's disease by combining serial analysis of gene expression and genetic linkage. Human Molecular Genetics, 2003, 12, 671-677.	2.9	17
138	A Radiation Hybrid Breakpoint Map of the Acute Myeloid Leukemia (AML) and Limb-Girdle Muscular Dystrophy 1A (LGMD1A) Regions of Chromosome 5q31 Localizing 122 Expressed Sequences. Genomics, 1999, 57, 24-35.	2.9	16
139	Haplotype Analysis in Icelandic Families Defines a Minimal Interval for the Macular Corneal Dystrophy Type I Gene. American Journal of Human Genetics, 1998, 63, 912-917.	6.2	15
140	Glutathione S-transferase polymorphisms and onset age in Î±-synuclein A53T mutant Parkinson's disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 254-258.	1.7	15
141	Charcot-Marie-Tooth Disease Type 2. Annals of the New York Academy of Sciences, 1999, 883, 42-46.	3.8	13
142	Fine mapping and genetic heterogeneity in the pure form of autosomal dominant familial spastic paraplegia. Neurogenetics, 2001, 3, 91-97.	1.4	13
143	High-Resolution Survey in Familial Parkinson Disease Genes Reveals Multiple Independent Copy Number Variation Events in PARK2. Human Mutation, 2013, 34, 1071-1074.	2.5	13
144	Knock-Down DHDDS Expression Induces Photoreceptor Degeneration in Zebrafish. Advances in Experimental Medicine and Biology, 2014, 801, 543-550.	1.6	13

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145	Confirmation of linkage of oculopharyngeal muscular dystrophy to chromosome 14q11.2-q13 in American families suggests the existence of a second causal mutation. Neuromuscular Disorders, 1997, 7, S75-S81.	0.6	12
146	The Q7R Saitohin gene polymorphism is not associated with Alzheimer disease. Neuroscience Letters, 2003, 347, 143-146.	2.1	12
147	Molecular Genetics of Autosomal-Dominant Axonal Charcot-Marie-Tooth Disease. NeuroMolecular Medicine, 2006, 8, 63-74.	3.4	12
148	Use of local genetic ancestry to assess TOMM40 -523â€² and risk for Alzheimer disease. Neurology: Genetics, 2020, 6, e404.	1.9	12
149	Identifying differential regulatory control of APOE É›4 on African versus European haplotypes as potential therapeutic targets. Alzheimer's and Dementia, 2022, 18, 1930-1942.	0.8	12
150	Statistical Viewer: a tool to upload and integrate linkage and association data as plots displayed within the Ensembl genome browser. BMC Bioinformatics, 2005, 6, 95.	2.6	11
151	Combinatorial Mismatch Scan (CMS) for loci associated with dementia in the Amish. BMC Medical Genetics, 2006, 7, 19.	2.1	11
152	Genome-wide Linkage Screen in Familial Parkinson Disease Identifies Loci on Chromosomes 3 and 18. American Journal of Human Genetics, 2009, 84, 499-504.	6.2	11
153	DNA variants in CACNA1C modify Parkinson disease risk only when vitamin D level is deficient. Neurology: Genetics, 2016, 2, e72.	1.9	11
154	hVMAT2: A Target of Individualized Medication for Parkinson's Disease. Neurotherapeutics, 2016, 13, 623-634.	4.4	11
155	Transcriptomic analysis of synovial extracellular RNA following knee trauma: A pilot study. Journal of Orthopaedic Research, 2018, 36, 1659-1665.	2.3	11
156	Dissecting the role of Amerindian genetic ancestry and the ApoE Îµ4 allele on Alzheimer disease in an admixed Peruvian population. Neurobiology of Aging, 2021, 101, 298.e11-298.e15.	3.1	11
157	Genomic Signatures of a Global Fitness Index in a Multiâ€Ethnic Cohort of Women. Annals of Human Genetics, 2013, 77, 147-157.	0.8	10
158	Motivations for Participation in Parkinson Disease Genetic Research Among Hispanics versus Non-Hispanics. Frontiers in Genetics, 2019, 10, 658.	2.3	10
159	The Puerto Rico Alzheimer Disease Initiative (PRADI): A Multisource Ascertainment Approach. Frontiers in Genetics, 2019, 10, 538.	2.3	10
160	Mutant C. elegans mitofusin leads to selective removal of mtDNA heteroplasmic deletions across generations to maintain fitness. BMC Biology, 2022, 20, 40.	3.8	9
161	A novel mutation in the von Hippel â€" Lindau gene. Human Molecular Genetics, 1994, 3, 1423-1424.	2.9	8
162	Reduction in the minimum candidate interval in the dominant-intermediate form of Charcot-Marie-Tooth neuropathy to D19S586 to D19S432. Neurogenetics, 2002, 4, 83-85.	1.4	8

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163	Lower Levels of Education Are Associated with Cognitive Impairment in the Old Order Amish. Journal of Alzheimer's Disease, 2021, 79, 451-458.	2.6	8
164	Derivation of stem cell line UMi028-A-2 containing a CRISPR/Cas9 induced Alzheimerâ€™s disease risk variant p.S1038C in the TTC3 gene. Stem Cell Research, 2021, 52, 102258.	0.7	7
165	Prenatal diagnosis using deletion studies in Duchenne muscular dystrophy. Prenatal Diagnosis, 1988, 8, 427-437.	2.3	6
166	Evidence for Genetic Heterogeneity Supports Clinical Differences in Congenital Myasthenic Syndromes. Human Heredity, 1998, 48, 325-332.	0.8	6
167	Genetic Polymorphisms Associated with Risk for Pulmonary Hypertension and Proteinuria in Sickle Cell Disease.. Blood, 2004, 104, 1668-1668.	1.4	6
168	Exclusion of Identified LGMD1 Loci from Four Dominant Limb-Girdle Muscular Dystrophy Families. Human Heredity, 1998, 48, 179-184.	0.8	5
169	Glutathione S-transferase omega-1 modifies age-at-onset of Alzheimer disease and Parkinson disease. Human Molecular Genetics, 2004, 13, 573-573.	2.9	5
170	Variants in chondroitin sulfate metabolism genes in thrombotic storm. Thrombosis Research, 2018, 161, 43-51.	1.7	5
171	Gabapentin Relieves Vertigo of Periodic Vestibulocerebellar Ataxia: 3 Cases and Possible Mechanism. Movement Disorders, 2021, 36, 1264-1267.	3.9	5
172	Clinical and Genetic Profiles of the Aging Sickle Cell Patient.. Blood, 2005, 106, 75-75.	1.4	5
173	Genomic Medicine and Neurology. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 249-267.	0.8	4
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175	Linkage of Alzheimer disease families with Puerto Rican ancestry identifies a chromosome 9 locus. Neurobiology of Aging, 2021, 104, 115.e1-115.e7.	3.1	4
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