Jeffery M Vance

List of Publications by Year in Descending Order

231 papers	18,420 citations	64 h-index	134 g-index
267 ext. papers	21,089 ext. citations	7.2 avg, IF	5.6 L-index

#	Paper	IF	Citations
231	Mutant C. elegans mitofusin leads to selective removal of mtDNA heteroplasmic deletions across generations to maintain fitness.. *BMC Biology*, **2022**, 20, 40	7.3	1
230	New insights into the genetic etiology of Alzheimer's disease and related dementias.. *Nature Genetics*, **2022**,	36.3	27
229	A novel duplication involving in a Turkish family supports its role in North Carolina macular dystrophy (NCMD/MCDR1). *Molecular Vision*, **2021**, 27, 518-527	2.3	0
228	Derivation of stem cell line UMi028-A-2 containing a CRISPR/Cas9 induced Alzheimer's disease risk variant p.S1038C in the TTC3 gene. *Stem Cell Research*, **2021**, 52, 102258	1.6	3
227	Genomewide Association Studies of LRRK2 Modifiers of Parkinson's Disease. *Annals of Neurology*, **2021**, 90, 76-88	9.4	9
226	Dissecting the role of Amerindian genetic ancestry and the ApoE ε4 allele on Alzheimer disease in an admixed Peruvian population. *Neurobiology of Aging*, **2021**, 101, 298.e11-298.e15	5.6	2
225	Gabapentin Relieves Vertigo of Periodic Vestibulocerebellar Ataxia: 3 Cases and Possible Mechanism. *Movement Disorders*, **2021**, 36, 1264-1267	7	2
224	Lower Levels of Education Are Associated with Cognitive Impairment in the Old Order Amish. *Journal of Alzheimerls Disease*, **2021**, 79, 451-458	4.3	0
223	Increased APOE ε4 expression is associated with the difference in Alzheimer's disease risk from diverse ancestral backgrounds. *Alzheimerls and Dementia*, **2021**, 17, 1179-1188	1.2	4
222	Successful Management of Catastrophic Thrombotic Storm in a Young Boy: A Case Report From Northern India. *Journal of Pediatric Hematology/Oncology*, **2021**, 43, e1132-e1135	1.2	1
221	Linkage of Alzheimer disease families with Puerto Rican ancestry identifies a chromosome 9 locus. *Neurobiology of Aging*, **2021**, 104, 115.e1-115.e7	5.6	0
220	Reply to: Gabapentin Relieves Vertigo of Periodic Vestibulocerebellar Ataxia: 3 Cases and Possible Mechanism. *Movement Disorders*, **2021**, 36, 1991	7	
219	ADSP follow-up study: NCRAD biospecimens.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056242	1.2	
218	Transgenic APOEε4/4 overexpression induces reactivity in astrocytes with a European APOEε4/4 local ancestry, but not in astrocytes with an African APOEε4/4 local ancestry.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056397	1.2	
217	Association of a locus on chromosome 17 with earlier age at onset of cognitive impairment in a familial Amish dataset.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056288	1.2	
216	Genome-wide association for protective variants in Alzheimer's disease in the Midwestern Amish.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056363	1.2	
215	Ancestry-specific intronic variants on the APOEe4 haplotype influence enhancer activity and interaction with APOE promoter.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e055266	1.2	

214	Preferential preservation of constructional praxis delayed recall compared to word list delayed recall in the Amish.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056386	1.2	
213	APOE-stratified genome-wide association analysis identifies novel Alzheimer disease candidate risk loci for African Americans.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056383	1.2	0
212	Genetic risk score for Alzheimer's disease in the Amish highlights differences in the genetic architecture compared to other European ancestry populations.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e053304	1.2	
211	Characterization of an Alzheimer disease-associated deletion in SORL1.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e055472	1.2	
210	ATAC-seq on iPSC derived astrocytes to assess chromatin accessibility differences between African and European local ancestry.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056086	1.2	
209	The Alzheimer's Disease Sequencing Project - Follow Up Study (ADSP-FUS): Increasing ethnic diversity in Alzheimer's genetics research with the addition of potential new cohorts.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056101	1.2	
208	African locus reduces the effect of ApoE 4 allele in Alzheimer's disease.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056210	1.2	
207	Expression quantitative trait loci (eQTL) analysis in a diverse Alzheimer disease cohort reveals ancestry-specific regulatory architectures.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056211	1.2	
206	Suggestive linkage and association of preserved cognition to chromosome 18 in genetically at-risk Amish.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056306	1.2	
205	Derivation of a CRISPR genome edited stem cell line containing a risk variant in TTC3.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056331	1.2	
204	Genome-wide association study of cognitive status and decline in the Amish.. *Alzheimerls and Dementia*, **2021**, 17 Suppl 3, e056525	1.2	
203	Longitudinal assessment of cognitive decline in the Amish. *Alzheimerls and Dementia*, **2020**, 16, e043440	1.2	
202	Recruitment strategies for the genetics of Alzheimer disease in the Puerto Rican population. *Alzheimerls and Dementia*, **2020**, 16, e043468	1.2	
201	Exploring the role of Amerindian genetic ancestry and ApoE4 gene on Alzheimer disease in the Peruvian population. *Alzheimerls and Dementia*, **2020**, 16, e045012	1.2	
200	Search for protective genetic variants in Alzheimer disease in the U.S. Midwestern Amish. *Alzheimerls and Dementia*, **2020**, 16, e045350	1.2	
199	A multiancestry analysis of Alzheimer disease coexpressed gene networks identifies a common immune signaling pathway regulated by granulocyte-colony stimulating factor (G-CSF). *Alzheimerls and Dementia*, **2020**, 16, e045361	1.2	
198	Increased APOE-e4 expression is associated with reactive A1 astrocytes and may confer the difference in Alzheimer disease risk from different ancestral backgrounds. *Alzheimerls and Dementia*, **2020**, 16, e045415	1.2	
197	African and European local ancestry surrounding Apolipoprotein E has a differential biological effect upon acute amyloid beta exposure in iPSC-differentiated astrocytes. *Alzheimerls and Dementia*, **2020**, 16, e045424	1.2	

196	Functional characterization of an Alzheimer disease-associated deletion in SORL1. *Alzheimerls and Dementia*, **2020**, 16, e045888	1.2	
195	Transcriptomic characterization of a Puerto Rican Alzheimer disease cohort implicates convergent immune-related pathways. *Alzheimerls and Dementia*, **2020**, 16, e045890	1.2	
194	Development of a massively parallel reporter assay to identify functional regulatory variants in the PICALM Alzheimer disease associated locus. *Alzheimerls and Dementia*, **2020**, 16, e045908	1.2	
193	Southern European genetic ancestry shows reduced APOE E4 risk for Alzheimer disease in Caribbean Hispanic population. *Alzheimerls and Dementia*, **2020**, 16, e045951	1.2	
192	Identification of differential regulation of European versus African local ancestry haplotypes surrounding ApoE4. *Alzheimerls and Dementia*, **2020**, 16, e046016	1.2	
191	Functional analysis of candidate genes identified through whole genome sequencing in Caribbean Hispanic families for late-onset Alzheimer disease. *Alzheimerls and Dementia*, **2020**, 16, e046017	1.2	0
190	The effect of global ancestry and diabetes on the 3MS score in older Puerto Ricans. *Alzheimerls and Dementia*, **2020**, 16, e046051	1.2	
189	Education and its effect on risk and age at onset in Alzheimer disease (AD) in African Americans. *Alzheimerls and Dementia*, **2020**, 16, e046078	1.2	
188	iPSC-derived neurons and microglia with an African-specific ABCA7 frameshift deletion have impaired function. *Alzheimerls and Dementia*, **2020**, 16, e046109	1.2	1
187	Recruiting African American males in Alzheimer's disease education and genetics research. *Alzheimerls and Dementia*, **2020**, 16, e046178	1.2	
186	The Alzheimer disease sequencing project follow up study (ADSP-FUS): Increasing ethnic diversity in Alzheimer genetics research with addition of potential new cohorts. *Alzheimerls and Dementia*, **2020**, 16, e046400	1.2	0
185	Joint linkage and association mapping of preserved cognition in the old-order Amish. *Alzheimerls and Dementia*, **2020**, 16, e046416	1.2	
184	PRADI cohort case-control study on related factors of Alzheimer disease. *Alzheimerls and Dementia*, **2020**, 16, e046443	1.2	
183	Use of local genetic ancestry to assess -523' and risk for Alzheimer disease. *Neurology: Genetics*, **2020**, 6, e404	3.8	7
182	RNA editing alterations in a multi-ethnic Alzheimer disease cohort converge on immune and endocytic molecular pathways. *Human Molecular Genetics*, **2019**, 28, 3053-3061	5.6	7
181	Motivations for Participation in Parkinson Disease Genetic Research Among Hispanics versus Non-Hispanics. *Frontiers in Genetics*, **2019**, 10, 658	4.5	5
180	Genome-wide brain DNA methylation analysis suggests epigenetic reprogramming in Parkinson disease. *Neurology: Genetics*, **2019**, 5, e342	3.8	27
179	The Puerto Rico Alzheimer Disease Initiative (PRADI): A Multisource Ascertainment Approach. *Frontiers in Genetics*, **2019**, 10, 538	4.5	3

178	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
177	Functional variants in the gene confer shared effects on risk for Crohn's disease and Parkinson's disease. *Science Translational Medicine*, **2018**, 10,	17.5	165
176	Variants in chondroitin sulfate metabolism genes in thrombotic storm. *Thrombosis Research*, **2018**, 161, 43-51	8.2	3
175	Transcriptomic analysis of synovial extracellular RNA following knee trauma: A pilot study. *Journal of Orthopaedic Research*, **2018**, 36, 1659-1665	3.8	9
174	P3-034: CONTINUOUS COMMUNITY ENGAGEMENT IMPROVES RECRUITMENT OF OLDER AFRICAN AMERICANS FOR GENETIC STUDIES IN ALZHEIMER'S DISEASE **2018**, 14, P1077-P1078		
173	P1-154: GENOME-WIDE LINKAGE ANALYSES OF AFRICAN AMERICAN FAMILIES SUPPORTS EVIDENCE OF LINKAGE TO CHROMOSOME 12 **2018**, 14, P336-P336		
172	O2-01-05: MULTI-ETHNIC ALZHEIMER'S DISEASE RELATED CHANGES OF RNA EDITING AFFECT IMMUNE REGULATION, ENDOCYTOSIS, AND AMYLOID PRECURSOR PROTEIN CATABOLISM **2018**, 14, P609-P610		
171	Ancestral origin of ApoE 4 Alzheimer disease risk in Puerto Rican and African American populations. *PLoS Genetics*, **2018**, 14, e1007791	6	56
170	Convergent Pathways in Idiopathic Autism Revealed by Time Course Transcriptomic Analysis of Patient-Derived Neurons. *Scientific Reports*, **2018**, 8, 8423	4.9	51
169	Genetic Variants of Microtubule Actin Cross-linking Factor 1 (MACF1) Confer Risk for Parkinson's Disease. *Molecular Neurobiology*, **2017**, 54, 2878-2888	6.2	16
168	Early-Onset Alzheimer Disease and Candidate Risk Genes Involved in Endolysosomal Transport. *JAMA Neurology*, **2017**, 74, 1113-1122	17.2	30
167	[P3169]: A PATIENT-DERIVED IPSC MODEL OF A RARE TTC3 MUTATION **2017**, 13, P999-P999		
166	[P2075]: INFLUENCE OF COMMUNITY ENGAGED FAMILY CONNECTOR IN RECRUITING AND ASCERTAINING AFRICAN AMERICANS FAMILY MEMBERS FOR GENOMIC RESEARCH **2017**, 13, P634-P635		
165	[P2102]: THE PUERTO RICO ALZHEIMER DISEASE INITIATIVE (PRADI): A MULTISOURCE ASCERTAINMENT APPROACH **2017**, 13, P646-P646		
164	[P2113]: THE RELEVANCE OF APOE4 TO ALZHEIMER's DISEASE IN THE PRESENCE OF LOCAL ANCESTRY DIFFERENCES. *Alzheimerls and Dementia*, **2017**, 13, P650	1.2	
163	Identification of TMEM230 mutations in familial Parkinson's disease. *Nature Genetics*, **2016**, 48, 733-9	36.3	122
162	ABCA7 frameshift deletion associated with Alzheimer disease in African Americans. *Neurology: Genetics*, **2016**, 2, e79	3.8	43
161	Overlap between Parkinson disease and Alzheimer disease in ABCA7 functional variants. *Neurology: Genetics*, **2016**, 2, e44	3.8	23

160	Regional Differential Genetic Response of Human Articular Cartilage to Impact Injury. *Cartilage*, **2016**, 7, 163-73	3	2
159	DNA variants in CACNA1C modify Parkinson disease risk only when vitamin D level is deficient. *Neurology: Genetics*, **2016**, 2, e72	3.8	10
158	hVMAT2: A Target of Individualized Medication for Parkinson's Disease. *Neurotherapeutics*, **2016**, 13, 623-34	6.4	8
157	Segregation of a rare TTC3 variant in an extended family with late-onset Alzheimer disease. *Neurology: Genetics*, **2016**, 2, e41	3.8	31
156	Vitamin D from different sources is inversely associated with Parkinson disease. *Movement Disorders*, **2015**, 30, 560-6	7	41
155	hVGAT-mCherry: A novel molecular tool for analysis of GABAergic neurons derived from human pluripotent stem cells. *Molecular and Cellular Neurosciences*, **2015**, 68, 244-57	4.8	16
154	PARK10 is a major locus for sporadic neuropathologically confirmed Parkinson disease. *Neurology*, **2015**, 84, 972-80	6.5	38
153	Detecting genetic interactions in pathway-based genome-wide association studies. *Genetic Epidemiology*, **2014**, 38, 300-9	2.6	12
152	Absence of C9ORF72 expanded or intermediate repeats in autopsy-confirmed Parkinson's disease. *Movement Disorders*, **2014**, 29, 827-30	7	20
151	Mutation K42E in dehydrodolichol diphosphate synthase (DHDDS) causes recessive retinitis pigmentosa. *Advances in Experimental Medicine and Biology*, **2014**, 801, 165-70	3.6	11
150	Knock-down DHDDS expression induces photoreceptor degeneration in zebrafish. *Advances in Experimental Medicine and Biology*, **2014**, 801, 543-50	3.6	11
149	C9ORF72 intermediate repeat copies are a significant risk factor for Parkinson disease. *Annals of Human Genetics*, **2013**, 77, 351-63	2.2	60
148	Genomic signatures of a global fitness index in a multi-ethnic cohort of women. *Annals of Human Genetics*, **2013**, 77, 147-57	2.2	7
147	A loss-of-function variant in the human histidyl-tRNA synthetase (HARS) gene is neurotoxic in vivo. *Human Mutation*, **2013**, 34, 191-9	4.7	85
146	High-resolution survey in familial Parkinson disease genes reveals multiple independent copy number variation events in PARK2. *Human Mutation*, **2013**, 34, 1071-4	4.7	12
145	Whole exome sequencing of rare variants in EIF4G1 and VPS35 in Parkinson disease. *Neurology*, **2013**, 80, 982-9	6.5	59
144	A recurrent loss-of-function alanyl-tRNA synthetase (AARS) mutation in patients with Charcot-Marie-Tooth disease type 2N (CMT2N). *Human Mutation*, **2012**, 33, 244-53	4.7	75
143	Clinical causes and treatment of the thrombotic storm. *Expert Review of Hematology*, **2012**, 5, 653-9	2.8	21

142	Derivation of autism spectrum disorder-specific induced pluripotent stem cells from peripheral blood mononuclear cells. *Neuroscience Letters*, **2012**, 516, 9-14	3.3	53
141	Notch activation induces endothelial cell senescence and pro-inflammatory response: implication of Notch signaling in atherosclerosis. *Atherosclerosis*, **2012**, 225, 296-303	3.1	77
140	Tyrosine hydroxylase gene: another piece of the genetic puzzle of Parkinson's disease. *CNS and Neurological Disorders - Drug Targets*, **2012**, 11, 469-81	2.6	13
139	Meta-analysis of Parkinson's disease: identification of a novel locus, RIT2. *Annals of Neurology*, **2012**, 71, 370-84	9.4	214
138	Gene expression profiles in Parkinson disease prefrontal cortex implicate FOXO1 and genes under its transcriptional regulation. *PLoS Genetics*, **2012**, 8, e1002794	6	54
137	Comprehensive research synopsis and systematic meta-analyses in Parkinson's disease genetics: The PDGene database. *PLoS Genetics*, **2012**, 8, e1002548	6	420
136	Mutations in the ER-shaping protein reticulon 2 cause the axon-degenerative disorder hereditary spastic paraplegia type 12. *Journal of Clinical Investigation*, **2012**, 122, 538-44	15.9	108
135	Identifying consensus disease pathways in Parkinson's disease using an integrative systems biology approach. *PLoS ONE*, **2011**, 6, e16917	3.7	58
134	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	6	163
133	Thrombotic storm revisited: preliminary diagnostic criteria suggested by the thrombotic storm study group. *American Journal of Medicine*, **2011**, 124, 290-6	2.4	33
132	Comparison of three targeted enrichment strategies on the SOLiD sequencing platform. *PLoS ONE*, **2011**, 6, e18595	3.7	60
131	Genomic medicine and neurology. *CONTINUUM Lifelong Learning in Neurology*, **2011**, 17, 249-67	3	3
130	Vitamin D receptor gene as a candidate gene for Parkinson disease. *Annals of Human Genetics*, **2011**, 75, 201-10	2.2	72
129	Whole-exome sequencing links a variant in DHDDS to retinitis pigmentosa. *American Journal of Human Genetics*, **2011**, 88, 201-6	11	130
128	Mutation screening of mitofusin 2 in Charcot-Marie-Tooth disease type 2. *Journal of Neurology*, **2011**, 258, 1234-9	5.5	30
127	A noise-reduction GWAS analysis implicates altered regulation of neurite outgrowth and guidance in autism. *Molecular Autism*, **2011**, 2, 1	6.5	139
126	Exome sequencing allows for rapid gene identification in a Charcot-Marie-Tooth family. *Annals of Neurology*, **2011**, 69, 464-70	9.4	91
125	Convergence of miRNA expression profiling, □synuclein interacton and GWAS in Parkinson's disease. *PLoS ONE*, **2011**, 6, e25443	3.7	184

124	Genome-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	2.2	374
123	Dementia revealed: novel chromosome 6 locus for late-onset Alzheimer disease provides genetic evidence for folate-pathway abnormalities. *PLoS Genetics*, **2010**, 6, e1001130	6	111
122	The transcription factor orthodenticle homeobox 2 influences axonal projections and vulnerability of midbrain dopaminergic neurons. *Brain*, **2010**, 133, 2022-31	11.2	38
121	Findings from a community education needs assessment to facilitate the integration of genomic medicine into primary care. *Genetics in Medicine*, **2010**, 12, 587-93	8.1	15
120	A potential novel variant of hereditary sensory neuropathy in a 61-year-old man with cough-induced syncope and vertebral artery dissection. *Mayo Clinic Proceedings*, **2010**, 85, 594-5	6.4	2
119	Gene-environment interactions in Parkinson's disease and other forms of parkinsonism. *NeuroToxicology*, **2010**, 31, 598-602	4.4	55
118	The mtDNA mutation spectrum of the progeroid Polg mutator mouse includes abundant control region multimers. *Cell Metabolism*, **2010**, 12, 675-82	24.6	71
117	PGC-1□a potential therapeutic target for early intervention in Parkinson's disease. *Science Translational Medicine*, **2010**, 2, 52ra73	17.5	546
116	Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. *Journal of Neurology*, **2010**, 257, 735-41	5.5	21
115	A rare novel deletion of the tyrosine hydroxylase gene in Parkinson disease. *Human Mutation*, **2010**, 31, E1767-71	4.7	22
114	SRRM2, a potential blood biomarker revealing high alternative splicing in Parkinson's disease. *PLoS ONE*, **2010**, 5, e9104	3.7	62
113	Neuropeptide Y gene polymorphisms confer risk of early-onset atherosclerosis. *PLoS Genetics*, **2009**, 5, e1000318	6	74
112	Genetic effects in the leukotriene biosynthesis pathway and association with atherosclerosis. *Human Genetics*, **2009**, 125, 217-29	6.3	44
111	Genome-wide linkage screen in familial Parkinson disease identifies loci on chromosomes 3 and 18. *American Journal of Human Genetics*, **2009**, 84, 499-504	11	8
110	Pesticide exposure and risk of Parkinson's disease: a family-based case-control study. *BMC Neurology*, **2008**, 8, 6	3.1	168
109	Comprehensive genetic analysis of the platelet activating factor acetylhydrolase (PLA2G7) gene and cardiovascular disease in case-control and family datasets. *Human Molecular Genetics*, **2008**, 17, 1318-28	5.6	62
108	Identification of genetic polymorphisms associated with risk for pulmonary hypertension in sickle cell disease. *Blood*, **2008**, 111, 5721-6	2.2	54
107	Nitric oxide synthase genes and their interactions with environmental factors in Parkinson's disease. *Neurogenetics*, **2008**, 9, 249-62	3	75

106	Variation in the miRNA-433 binding site of FGF20 confers risk for Parkinson disease by overexpression of alpha-synuclein. *American Journal of Human Genetics*, **2008**, 82, 283-9	11	403
105	Response to Zaykin and Shibata. *American Journal of Human Genetics*, **2008**, 82, 796-797	11	16
104	Glutathione S-transferase polymorphisms and onset age in alpha-synuclein A53T mutant Parkinson's disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2007**, 144B, 254-8	3.5	12
103	Genetic polymorphisms associated with priapism in sickle cell disease. *British Journal of Haematology*, **2007**, 137, 262-7	4.5	54
102	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, 955-60	11.5	381
101	Smoking, caffeine, and nonsteroidal anti-inflammatory drugs in families with Parkinson disease. *Archives of Neurology*, **2007**, 64, 576-80		96
100	No gene is an island: the flip-flop phenomenon. *American Journal of Human Genetics*, **2007**, 80, 531-8	11	402
99	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, 650-63	11	96
98	Neurology and Genomic Medicine **2007**, 19-28		
97	Family-based case-control study of MAOA and MAOB polymorphisms in Parkinson disease. *Movement Disorders*, **2006**, 21, 2175-80	7	29
96	A genome-wide linkage analysis of dementia in the Amish. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2006**, 141B, 160-6	3.5	35
95	Axonal neuropathy with optic atrophy is caused by mutations in mitofusin 2. *Annals of Neurology*, **2006**, 59, 276-81	9.4	276
94	MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. *Brain*, **2006**, 129, 2093-102	11.2	295
93	GATA2 is associated with familial early-onset coronary artery disease. *PLoS Genetics*, **2006**, 2, e139	6	71
92	Mechanisms of disease: a molecular genetic update on hereditary axonal neuropathies. *Nature Clinical Practice Neurology*, **2006**, 2, 45-53		74
91	Mutations in the novel mitochondrial protein REEP1 cause hereditary spastic paraplegia type 31. *American Journal of Human Genetics*, **2006**, 79, 365-9	11	181
90	Molecular genetics of autosomal-dominant axonal Charcot-Marie-Tooth disease. *NeuroMolecular Medicine*, **2006**, 8, 63-74	4.6	62
89	Revealing the role of glutathione S-transferase omega in age-at-onset of Alzheimer and Parkinson diseases. *Neurobiology of Aging*, **2006**, 27, 1087-93	5.6	53

88	Combinatorial Mismatch Scan (CMS) for loci associated with dementia in the Amish. *BMC Medical Genetics*, **2006**, 7, 19	2.1	7
87	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, 307-16	5.3	36
86	Molecular Genetics of Autosomal-Dominant Axonal Charcot-Marie-Tooth Disease. *NeuroMolecular Medicine*, **2006**, 8, 63-74	4.6	9
85	NOS2A and the modulating effect of cigarette smoking in Parkinson's disease. *Annals of Neurology*, **2006**, 60, 366-73	9.4	31
84	Identification of risk and age-at-onset genes on chromosome 1p in Parkinson disease. *American Journal of Human Genetics*, **2005**, 77, 252-64	11	53
83	SNPselector: a web tool for selecting SNPs for genetic association studies. *Bioinformatics*, **2005**, 21, 4181-6	7.6	95
82	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, 1118-20	4.9	14
81	Mutations in the pleckstrin homology domain of dynamin 2 cause dominant intermediate Charcot-Marie-Tooth disease. *Nature Genetics*, **2005**, 37, 289-94	36.3	278
80	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	3.6	10
79	Genomic convergence to identify candidate genes for Parkinson disease: SAGE analysis of the substantia nigra. *Movement Disorders*, **2005**, 20, 1299-309	7	35
78	Emerging pathways for hereditary axonopathies. *Journal of Molecular Medicine*, **2005**, 83, 935-43	5.5	27
77	Association between the neuron-specific RNA-binding protein ELAVL4 and Parkinson disease. *Human Genetics*, **2005**, 117, 27-33	6.3	58
76	Expression profiling of substantia nigra in Parkinson disease, progressive supranuclear palsy, and frontotemporal dementia with parkinsonism. *Archives of Neurology*, **2005**, 62, 917-21		128
75	A mutation in the TRPC6 cation channel causes familial focal segmental glomerulosclerosis. *Science*, **2005**, 308, 1801-4	33.3	829
74	Clinical and Genetic Profiles of the Aging Sickle Cell Patient.. *Blood*, **2005**, 106, 75-75	2.2	4
73	Priapism in SCD: Clinical and Genetic Correlations.. *Blood*, **2005**, 106, 3174-3174	2.2	
72	Glutathione S-transferase omega-1 modifies age-at-onset of Alzheimer disease and Parkinson disease. *Human Molecular Genetics*, **2004**, 13, 573-573	5.6	4
71	Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. *Nature Genetics*, **2004**, 36, 449-51	36.3	1243

70	Linkage disequilibrium and haplotype tagging polymorphisms in the Tau H1 haplotype. *Neurogenetics*, **2004**, 5, 147-55	3	22
69	Comprehensive association analysis of APOE regulatory region polymorphisms in Alzheimer disease. *Neurogenetics*, **2004**, 5, 201-8	3	21
68	Analysis of European mitochondrial haplogroups with Alzheimer disease risk. *Neuroscience Letters*, **2004**, 365, 28-32	3.3	237
67	Fibroblast growth factor 20 polymorphisms and haplotypes strongly influence risk of Parkinson disease. *American Journal of Human Genetics*, **2004**, 74, 1121-7	11	124
66	A genomewide scan for early-onset coronary artery disease in 438 families: the GENECARD Study. *American Journal of Human Genetics*, **2004**, 75, 436-47	11	132
65	Genetic Polymorphisms Associated with Risk for Pulmonary Hypertension and Proteinuria in Sickle Cell Disease.. *Blood*, **2004**, 104, 1668-1668	2.2	6
64	Association study of Parkin gene polymorphisms with idiopathic Parkinson disease. *Archives of Neurology*, **2003**, 60, 975-80		44
63	Parkin mutations and susceptibility alleles in late-onset Parkinson's disease. *Annals of Neurology*, **2003**, 53, 624-9	9.4	172
62	The deacetylase HDAC6 regulates aggresome formation and cell viability in response to misfolded protein stress. *Cell*, **2003**, 115, 727-38	56.2	1187
61	The Q7R Saitohin gene polymorphism is not associated with Alzheimer disease. *Neuroscience Letters*, **2003**, 347, 143-6	3.3	11
60	Mitochondrial polymorphisms significantly reduce the risk of Parkinson disease. *American Journal of Human Genetics*, **2003**, 72, 804-11	11	458
59	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, 1293-9	11	437
58	Ordered-subsets linkage analysis detects novel Alzheimer disease loci on chromosomes 2q34 and 15q22. *American Journal of Human Genetics*, **2003**, 73, 1041-51	11	87
57	Identification of MeCP2 mutations in a series of females with autistic disorder. *Pediatric Neurology*, **2003**, 28, 205-11	2.9	175
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