## Jonathan L Haines

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4479208/publications.pdf

Version: 2024-02-01

701 517 82,612 549 121 267 citations h-index g-index papers 613 613 613 69301 docs citations times ranked citing authors all docs

#	Article	IF	Citations
1	Finding the missing heritability of complex diseases. Nature, 2009, 461, 747-753.	27.8	7,490
2	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. Nature, 1993, 362, 59-62.	27.8	6,331
3	Meta-analysis of 74,046 individuals identifies $11$ new susceptibility loci for Alzheimer's disease. Nature Genetics, 2013, 45, 1452-1458.	21.4	3,741
4	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. Nature, 2011, 476, 214-219.	27.8	2,400
5	Complement Factor H Variant Increases the Risk of Age-Related Macular Degeneration. Science, 2005, 308, 419-421.	12.6	2,232
6	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	21.4	2,067
7	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates Aβ, tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	21.4	1,962
8	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
9	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. Nature Genetics, 2011, 43, 436-441.	21.4	1,676
10	Risk Alleles for Multiple Sclerosis Identified by a Genomewide Study. New England Journal of Medicine, 2007, 357, 851-862.	27.0	1,529
11	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
12	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. Nature Genetics, 2013, 45, 1353-1360.	21.4	1,213
13	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	21.4	1,167
14	Mutations in UBQLN2 cause dominant X-linked juvenile and adult-onset ALS and ALS/dementia. Nature, 2011, 477, 211-215.	27.8	1,016
15	Common genetic variants on $5p14.1$ associate with autism spectrum disorders. Nature, 2009, 459, 528-533.	27.8	912
16	Systematic comparison of phenome-wide association study of electronic medical record data and genome-wide association study data. Nature Biotechnology, 2013, 31, 1102-1111.	17.5	846
17	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	6.2	819
18	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	21.4	783

#	Article	IF	CITATIONS
19	Meta-analysis of genome scans and replication identify CD6, IRF8 and TNFRSF1A as new multiple sclerosis susceptibility loci. Nature Genetics, 2009, 41, 776-782.	21.4	729
20	Multiple sclerosis genomic map implicates peripheral immune cells and microglia in susceptibility. Science, 2019, 365, .	12.6	710
21	New insights into the genetic etiology of Alzheimer's disease and related dementias. Nature Genetics, 2022, 54, 412-436.	21.4	700
22	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	21.4	687
23	The Electronic Medical Records and Genomics (eMERGE) Network: past, present, and future. Genetics in Medicine, 2013, 15, 761-771.	2.4	611
24	The PhenX Toolkit: Get the Most From Your Measures. American Journal of Epidemiology, 2011, 174, 253-260.	3.4	610
25	Heterozygosity for a Surfactant Protein C Gene Mutation Associated with Usual Interstitial Pneumonitis and Cellular Nonspecific Interstitial Pneumonitis in One Kindred. American Journal of Respiratory and Critical Care Medicine, 2002, 165, 1322-1328.	5.6	597
26	Interleukin 7 receptor $\hat{l}_{\pm}$ chain ( IL7R ) shows allelic and functional association with multiple sclerosis. Nature Genetics, 2007, 39, 1083-1091.	21.4	578
27	Isolation of a novel gene underlying batten disease, CLN3. Cell, 1995, 82, 949-957.	28.9	554
28	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
29	Mitochondrial Polymorphisms Significantly Reduce the Risk of Parkinson Disease. American Journal of Human Genetics, 2003, 72, 804-811.	6.2	507
30	Genome-wide association study identifies a new breast cancer susceptibility locus at 6q25.1. Nature Genetics, 2009, 41, 324-328.	21.4	481
31	Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. Nature, 1987, 329, 246-248.	27.8	478
32	Genetic variants near <i>TIMP3</i> and high-density lipoprotein–associated loci influence susceptibility to age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7401-7406.	7.1	475
33	Genomeâ€Wide Association Study Confirms SNPs in <i>SNCA</i> and the <i>MAPT</i> Region as Common Risk Factors for Parkinson Disease. Annals of Human Genetics, 2010, 74, 97-109.	0.8	417
34	Linkage of a Gene Causing Familial Amyotrophic Lateral Sclerosis to Chromosome 21 and Evidence of Genetic-Locus Heterogeneity. New England Journal of Medicine, 1991, 324, 1381-1384.	27.0	407
35	Meta-analysis Confirms CR1, CLU, and PICALM as Alzheimer Disease Risk Loci and Reveals Interactions With APOE Genotypes. Archives of Neurology, 2010, 67, 1473.	4.5	376
36	Variants in the ATP-Binding Cassette Transporter (ABCA7), Apolipoprotein E $\ddot{l}\mu$ 4, and the Risk of Late-Onset Alzheimer Disease in African Americans. JAMA - Journal of the American Medical Association, 2013, 309, 1483.	7.4	360

#	Article	IF	CITATIONS
37	GWAS of Cerebrospinal Fluid Tau Levels Identifies Risk Variants for Alzheimer's Disease. Neuron, 2013, 78, 256-268.	8.1	344
38	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
39	A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. Nature Neuroscience, 2017, 20, 1052-1061.	14.8	330
40	Cigarette Smoking Strongly Modifies the Association of LOC387715 and Age-Related Macular Degeneration. American Journal of Human Genetics, 2006, 78, 852-864.	6.2	316
41	Genomeâ€wide metaâ€analysis identifies novel multiple sclerosis susceptibility loci. Annals of Neurology, 2011, 70, 897-912.	5.3	314
42	Class II HLA interactions modulate genetic risk for multiple sclerosis. Nature Genetics, 2015, 47, 1107-1113.	21.4	312
43	Mapping Multiple Sclerosis Susceptibility to the HLA-DR Locus in African Americans. American Journal of Human Genetics, 2004, 74, 160-167.	6.2	311
44	Genetic assessment of age-associated Alzheimer disease risk: Development and validation of a polygenic hazard score. PLoS Medicine, 2017, 14, e1002258.	8.4	311
45	Genome-Wide Association Meta-analysis of Neuropathologic Features of Alzheimer's Disease and Related Dementias. PLoS Genetics, 2014, 10, e1004606.	3.5	305
46	Common genetic variants in the CLDN2 and PRSS1-PRSS2 loci alter risk for alcohol-related and sporadic pancreatitis. Nature Genetics, 2012, 44, 1349-1354.	21.4	303
47	Robust Replication of Genotype-Phenotype Associations across Multiple Diseases in an Electronic Medical Record. American Journal of Human Genetics, 2010, 86, 560-572.	6.2	302
48	Cardiac Sodium Channel ( <i>SCN5A</i> ) Variants Associated with Atrial Fibrillation. Circulation, 2008, 117, 1927-1935.	1.6	292
49	Age at Onset in Two Common Neurodegenerative Diseases Is Genetically Controlled. American Journal of Human Genetics, 2002, 70, 985-993.	6.2	291
50	PheKB: a catalog and workflow for creating electronic phenotype algorithms for transportability. Journal of the American Medical Informatics Association: JAMIA, 2016, 23, 1046-1052.	4.4	284
51	Heterogeneity at the HLA-DRB1 locus and risk for multiple sclerosis. Human Molecular Genetics, 2006, 15, 2813-2824.	2.9	279
52	Common Variants at 9p21 and 8q22 Are Associated with Increased Susceptibility to Optic Nerve Degeneration in Glaucoma. PLoS Genetics, 2012, 8, e1002654.	3.5	276
53	The genetic defect in familial Alzheimer's disease is not tightly linked to the amyloid $\hat{l}^2$ -protein gene. Nature, 1987, 329, 156-157.	27.8	275
54	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. Nature Genetics, 2013, 45, 155-163.	21.4	269

#	Article	IF	CITATIONS
55	A High-Density Screen for Linkage in Multiple Sclerosis. American Journal of Human Genetics, 2005, 77, 454-467.	6.2	268
56	Analysis of European mitochondrial haplogroups with Alzheimer disease risk. Neuroscience Letters, 2004, 365, 28-32.	2.1	264
57	Quality Control Procedures for Genomeâ€Wide Association Studies. Current Protocols in Human Genetics, 2011, 68, Unit1.19.	3.5	259
58	Complete Genomic Screen in Parkinson Disease. JAMA - Journal of the American Medical Association, 2001, 286, 2239.	7.4	257
59	Temperature-sensitive mutations in the III–IV cytoplasmic loop region of the skeletal muscle sodium channel gene in paramyotonia congenita. Cell, 1992, 68, 769-774.	28.9	249
60	Human gene for torsion dystonia located on chromosome 9q32-q34. Neuron, 1989, 2, 1427-1434.	8.1	246
61	Exome sequencing of extended families with autism reveals genes shared across neurodevelopmental and neuropsychiatric disorders. Molecular Autism, 2014, 5, 1.	4.9	246
62	Exceptionally low likelihood of Alzheimer's dementia in APOE2 homozygotes from a 5,000-person neuropathological study. Nature Communications, 2020, 11, 667.	12.8	246
63	Genome-wide Association Study Implicates a Chromosome 12 Risk Locus for Late-Onset Alzheimer Disease. American Journal of Human Genetics, 2009, 84, 35-43.	6.2	242
64	Genetics, statistics and human disease: analytical retooling for complexity. Trends in Genetics, 2004, 20, 640-647.	6.7	230
65	Brain Expression Genome-Wide Association Study (eGWAS) Identifies Human Disease-Associated Variants. PLoS Genetics, 2012, 8, e1002707.	3.5	225
66	Meta-analysis of genome scans of age-related macular degeneration. Human Molecular Genetics, 2005, 14, 2257-2264.	2.9	224
67	Sex-Specific Association of Apolipoprotein E With Cerebrospinal Fluid Levels of Tau. JAMA Neurology, 2018, 75, 989.	9.0	223
68	Linkage of recessive familial amyotrophic lateral sclerosis to chromosome 2q33–q35. Nature Genetics, 1994, 7, 425-428.	21.4	221
69	Genome-wide analyses identify 68 new loci associated with intraocular pressure and improve risk prediction for primary open-angle glaucoma. Nature Genetics, 2018, 50, 778-782.	21.4	214
70	Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. Nature Genetics, 2014, 46, 1126-1130.	21.4	212
71	Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. Nature Genetics, 2016, 48, 189-194.	21.4	211
72	Glutathione S-transferase omega-1 modifiesage-at-onset of Alzheimer disease and Parkinson disease. Human Molecular Genetics, 2003, 12, 3259-3267.	2.9	208

#	Article	IF	Citations
73	A Genomeâ€wide Association Study of Autism Reveals a Common Novel Risk Locus at 5p14.1. Annals of Human Genetics, 2009, 73, 263-273.	0.8	207
74	Neonatal Pulmonary Hypertension. New England Journal of Medicine, 2001, 344, 1832-1838.	27.0	202
75	Parkin mutations and susceptibility alleles in lateâ€onset Parkinson's disease. Annals of Neurology, 2003, 53, 624-629.	5.3	201
76	Mutations in a Novel CLN6-Encoded Transmembrane Protein Cause Variant Neuronal Ceroid Lipofuscinosis in Man and Mouse. American Journal of Human Genetics, 2002, 70, 324-335.	6.2	199
77	Genome-wide association study identifies four novel loci associated with Alzheimer's endophenotypes and disease modifiers. Acta Neuropathologica, 2017, 133, 839-856.	7.7	199
78	Evidence for a role of the rare p.A152T variant in MAPT in increasing the risk for FTD-spectrum and Alzheimer's diseases. Human Molecular Genetics, 2012, 21, 3500-3512.	2.9	198
79	Prevalence of Mutations in TIGR/Myocilin in Patients with Adult and Juvenile Primary Open-Angle Glaucoma. American Journal of Human Genetics, 1998, 63, 1549-1552.	6.2	197
80	Genome-wide meta-analysis identifies 127 open-angle glaucoma loci with consistent effect across ancestries. Nature Communications, 2021, 12, 1258.	12.8	196
81	Clustering of autoimmune diseases in families with a high-risk for multiple sclerosis: a descriptive study. Lancet Neurology, The, 2006, 5, 924-931.	10.2	194
82	Incorporating language phenotypes strengthens evidence of linkage to autism. American Journal of Medical Genetics Part A, 2001, 105, 539-547.	2.4	192
83	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. Nature Genetics, 2020, 52, 160-166.	21.4	192
84	A noise-reduction GWAS analysis implicates altered regulation of neurite outgrowth and guidance in autism. Molecular Autism, $2011, 2, 1$ .	4.9	191
85	Whole exome sequencing study identifies novel rare and common Alzheimer's-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	7.9	191
86	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. Nature Genetics, 2014, 46, 1120-1125.	21.4	186
87	The role of the <i>CD58</i> locus in multiple sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 5264-5269.	7.1	185
88	Construction of a GT polymorphism map of human 9q. Genomics, 1992, 12, 229-240.	2.9	181
89	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. Human Genetics, 2012, 131, 565-579.	3.8	180
90	Functional Candidate Genes in Age-Related Macular Degeneration: Significant Association with <i>VEGF</i> , <i>VLDLR</i> , and <i>LRP6</i> , 2006, 47, 329.		178

#	Article	IF	Citations
91	Protective effect of complement factor B and complement component 2 variants in age-related macular degeneration. Human Molecular Genetics, 2007, 16, 1986-1992.	2.9	175
92	Age-Related Maculopathy: A Genomewide Scan with Continued Evidence of Susceptibility Loci within the 1q31, 10q26, and 17q25 Regions. American Journal of Human Genetics, 2004, 75, 174-189.	6.2	174
93	Assessment of the genetic variance of late-onset Alzheimer's disease. Neurobiology of Aging, 2016, 41, 200.e13-200.e20.	3.1	174
94	Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671.	0.8	173
95	Genome- and Phenome-Wide Analyses of Cardiac Conduction Identifies Markers of Arrhythmia Risk. Circulation, 2013, 127, 1377-1385.	1.6	167
96	Effects of Multiple Genetic Loci on Age at Onset in Late-Onset Alzheimer Disease. JAMA Neurology, 2014, 71, 1394.	9.0	166
97	Rapidly progressive Alzheimer's disease features distinct structures of amyloid-β. Brain, 2015, 138, 1009-1022.	7.6	166
98	Transethnic genomeâ€wide scan identifies novel Alzheimer's disease loci. Alzheimer's and Dementia, 2017, 13, 727-738.	0.8	166
99	Evidence for Polygenic Susceptibility to Multiple Sclerosisâ€"The Shape of Things to Come. American Journal of Human Genetics, 2010, 86, 621-625.	6.2	162
100	Neurofibromatosis 2: Clinical and DNA Linkage Studies of a Large Kindred. New England Journal of Medicine, 1988, 319, 278-283.	27.0	161
101	The Next PAGE in Understanding Complex Traits: Design for the Analysis of Population Architecture Using Genetics and Epidemiology (PAGE) Study. American Journal of Epidemiology, 2011, 174, 849-859.	3.4	161
102	A highly polymorphic locus very tightly linked to the Huntington's disease gene. Nature, 1988, 332, 734-736.	27.8	159
103	Evaluation of copy number variations reveals novel candidate genes in autism spectrum disorder-associated pathways. Human Molecular Genetics, 2012, 21, 3513-3523.	2.9	158
104	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. Nature Genetics, 2013, 45, 1375-1379.	21.4	158
105	A second major histocompatibility complex susceptibility locus for multiple sclerosis. Annals of Neurology, 2007, 61, 228-236.	5.3	156
106	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma in Caucasians from the USA. Human Molecular Genetics, 2011, 20, 4707-4713.	2.9	156
107	Whole-Exome Sequencing Links a Variant in DHDDS to Retinitis Pigmentosa. American Journal of Human Genetics, 2011, 88, 201-206.	6.2	155
108	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	2.5	155

#	Article	IF	CITATIONS
109	Linkage Disequilibrium at the Angelman Syndrome Gene UBE3A in Autism Families. Genomics, 2001, 77, 105-113.	2.9	154
110	Meta-analysis of the Association Between Variants in SORL1 and Alzheimer Disease. Archives of Neurology, 2011, 68, 99.	4.5	153
111	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
112	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. Nature Genetics, 1993, 4, 160-164.	21.4	149
113	SORL1 Is Genetically Associated with Late-Onset Alzheimer's Disease in Japanese, Koreans and Caucasians. PLoS ONE, 2013, 8, e58618.	2.5	149
114	Polygenic Overlap Between C-Reactive Protein, Plasma Lipids, and Alzheimer Disease. Circulation, 2015, 131, 2061-2069.	1.6	145
115	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. Neurology, 2012, 79, 221-228.	1.1	144
116	Novel Alzheimer Disease Risk Loci and Pathways in African American Individuals Using the African Genome Resources Panel. JAMA Neurology, 2021, 78, 102.	9.0	144
117	Rapid Communication: Cu/Zn Superoxide Dismutase Activity in Familial and Sporadic Amyotrophic Lateral Sclerosis. Journal of Neurochemistry, 1994, 62, 384-387.	3.9	143
118	Investigation of autism and GABA receptor subunit genes in multiple ethnic groups. Neurogenetics, 2006, 7, 167-174.	1.4	141
119	Common variants in Alzheimer's disease and risk stratification by polygenic risk scores. Nature Communications, 2021, 12, 3417.	12.8	140
120	A pooled case-control study of the apolipoprotein E (APOE) gene in age-related maculopathy. Ophthalmic Genetics, 2002, 23, 209-223.	1.2	136
121	A linkage disequilibrium map of the 1â€Mb 15q12 GABA <sub>A</sub> receptor subunit cluster and association to autism. American Journal of Medical Genetics Part A, 2004, 131B, 51-59.	2.4	135
122	Localization of the huntington's disease gene to a small segment of chromosome 4 flanked by D4S10 and the telomere. Cell, 1987, 50, 565-571.	28.9	130
123	Genome-wide and Ordered-Subset linkage analyses provide support for autism loci on $17q$ and $19p$ with evidence of phenotypic and interlocus genetic correlates. BMC Medical Genetics, $2005$ , $6$ , $1$ .	2.1	130
124	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
125	Recurrent Tissue-Specific mtDNA Mutations Are Common in Humans. PLoS Genetics, 2013, 9, e1003929.	3.5	130
126	Imputation and quality control steps for combining multiple genome-wide datasets. Frontiers in Genetics, 2014, 5, 370.	2.3	130

#	Article	IF	Citations
127	Mitochondrial haplogroups and peripheral neuropathy during antiretroviral therapy: an adult AIDS clinical trials group study. Aids, 2005, 19, 1341-1349.	2.2	129
128	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
129	Genetic susceptibility to Alzheimer disease. Trends in Genetics, 1995, 11, 504-508.	6.7	120
130	C3 R102G polymorphism increases risk of age-related macular degeneration. Human Molecular Genetics, 2008, 17, 1821-1824.	2.9	120
131	New insights into the genetics of primary open-angle glaucoma based on meta-analyses of intraocular pressure and optic disc characteristics Human Molecular Genetics, 2017, 26, ddw399.	2.9	120
132	Identification of Two Novel Loci for Dominantly Inherited Familial Amyotrophic Lateral Sclerosis. American Journal of Human Genetics, 2003, 73, 397-403.	6.2	119
133	Identification of Genomic Predictors of Atrioventricular Conduction. Circulation, 2010, 122, 2016-2021.	1.6	117
134	Ancestral origin of ApoE $\hat{l}\mu4$ Alzheimer disease risk in Puerto Rican and African American populations. PLoS Genetics, 2018, 14, e1007791.	3 <b>.</b> 5	117
135	A rare mutation in UNC5C predisposes to late-onset Alzheimer's disease and increases neuronal cell death. Nature Medicine, 2014, 20, 1452-1457.	30.7	116
136	An $\hat{1}\pm$ -2-macroglobulin insertion-deletion polymorphism in Alzheimer disease. Nature Genetics, 1999, 22, 19-21.	21.4	115
137	Replication analysis identifies TYK2 as a multiple sclerosis susceptibility factor. European Journal of Human Genetics, 2009, 17, 1309-1313.	2.8	115
138	Low-Frequency and Rare-Coding Variation Contributes to Multiple Sclerosis Risk. Cell, 2018, 175, 1679-1687.e7.	28.9	115
139	Distribution of WDR36DNA Sequence Variants in Patients with Primary Open-Angle Glaucoma., 2006, 47, 2542.		114
140	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. Nature Genetics, 2017, 49, 993-1004.	21.4	114
141	Exploratory Subsetting of Autism Families Based on Savant Skills Improves Evidence of Genetic Linkage to 15q11-q13. Journal of the American Academy of Child and Adolescent Psychiatry, 2003, 42, 856-863.	0.5	112
142	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. Human Molecular Genetics, 2018, 27, 1486-1496.	2.9	111
143	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. Annals of Neurology, 1990, 28, 614-621.	5 <b>.</b> 3	110
144	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

#	Article	IF	Citations
145	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
146	Genetic Variants Associated with Optic Nerve Vertical Cup-to-Disc Ratio Are Risk Factors for Primary Open Angle Glaucoma in a US Caucasian Population. , 2011, 52, 1788.		109
147	Comprehensive follow-up of the first genome-wide association study of multiple sclerosis identifies KIF21B and TMEM39A as susceptibility loci. Human Molecular Genetics, 2010, 19, 953-962.	2.9	108
148	Endothelial Nitric Oxide Synthase Gene Variants and Primary Open-Angle Glaucoma: Interactions with Sex and Postmenopausal Hormone Use., 2010, 51, 971.		107
149	Association of MAPT haplotypes with Alzheimer's disease risk and MAPT brain gene expression levels. Alzheimer's Research and Therapy, 2014, 6, 39.	6.2	106
150	DNA sequence variants in the LOXL1 gene are associated with pseudoexfoliation glaucoma in a U.S. clinic-based population with broad ethnic diversity. BMC Medical Genetics, 2008, 9, 5.	2.1	105
151	A common variant near TGFBR3 is associated with primary open angle glaucoma. Human Molecular Genetics, 2015, 24, 3880-3892.	2.9	105
152	SORCS1 alters amyloid precursor protein processing and variants may increase Alzheimer's disease risk. Annals of Neurology, 2011, 69, 47-64.	5.3	104
153	Dissecting the genetic relationship between cardiovascular risk factors and Alzheimer's disease. Acta Neuropathologica, 2019, 137, 209-226.	7.7	100
154	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
155	Mitochondrial DNA Polymorphism A4917G Is Independently Associated with Age-Related Macular Degeneration. PLoS ONE, 2008, 3, e2091.	2.5	99
156	Association Between Apolipoprotein E Genotype and Alzheimer Disease in African American Subjects. Archives of Neurology, 2002, 59, 594.	4.5	98
157	Comprehensive Search for Alzheimer Disease Susceptibility Loci in the APOE Region. Archives of Neurology, 2012, 69, 1270.	4.5	97
158	Mapping of the Disease Locus and Identification of ADAMTS10 As a Candidate Gene in a Canine Model of Primary Open Angle Glaucoma. PLoS Genetics, 2011, 7, e1001306.	3.5	96
159	Genetic Linkage of Autosomal Dominant Juvenile Glaucoma to 1q21-q31 in Three Affected Pedigrees. Genomics, 1994, 21, 299-303.	2.9	95
160	Assessing the accuracy of observer-reported ancestry in a biorepository linked to electronic medical records. Genetics in Medicine, 2010, 12, 648-650.	2.4	94
161	Two rare <i>AKAP9</i> variants are associated with Alzheimer's disease in African Americans. Alzheimer's and Dementia, 2014, 10, 609.	0.8	94
162	Recombination events suggest potential sites for the Huntington's disease gene. Neuron, 1989, 3, 183-190.	8.1	93

#	Article	IF	Citations
163	Genome-wide association study and meta-analysis of intraocular pressure. Human Genetics, 2014, 133, 41-57.	3.8	93
164	Genetic variants and functional pathways associated with resilience to Alzheimer's disease. Brain, 2020, 143, 2561-2575.	7.6	93
165	Analysis of the stargardt disease gene (ABCR) in age-related macular degenerationâ~†. Ophthalmology, 1999, 106, 1531-1536.	5.2	91
166	Drug Transporter and Metabolizing Enzyme Gene Variants and Nonnucleoside Reverse-Transcriptase Inhibitor Hepatotoxicity. Clinical Infectious Diseases, 2006, 43, 779-782.	5.8	91
167	Association of CAV1/CAV2 Genomic Variants with Primary Open-Angle Glaucoma Overall and by Gender and Pattern of Visual Field Loss. Ophthalmology, 2014, 121, 508-516.	5.2	91
168	Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. Nature Communications, 2014, 5, 4883.	12.8	89
169	Apolipoprotein E genotype in patients with alzheimer's disease: Implications for the risk of dementia among relatives. Annals of Neurology, 1995, 38, 797-808.	5.3	87
170	Two novel loci, <i>COBL</i> and <i>SLC10A2</i> , for Alzheimer's disease in African Americans. Alzheimer's and Dementia, 2017, 13, 119-129.	0.8	87
171	Sex-specific genetic predictors of Alzheimer's disease biomarkers. Acta Neuropathologica, 2018, 136, 857-872.	7.7	87
172	Lack of Association of Mutations in Optineurin With Disease in Patients With Adult-onset Primary Open-angle Glaucoma. JAMA Ophthalmology, 2003, 121, 1181.	2.4	86
173	Early Adult-Onset POAG Linked to 15q11-13 Using Ordered Subset Analysis. , 2005, 46, 2002.		86
174	Deletion of CFHR3 and CFHR1 genes in age-related macular degeneration. Human Molecular Genetics, 2008, 17, 971-977.	2.9	85
175	Localization of Age-Related Macular Degeneration-Associated ARMS2 in Cytosol, Not Mitochondria. , 2009, 50, 3084.		85
176	Distribution of Optineurin Sequence Variations in an Ethnically Diverse Population of Low-tension Glaucoma Patients From the United States. Journal of Glaucoma, 2006, 15, 358-363.	1.6	82
177	GATA2 Is Associated with Familial Early-Onset Coronary Artery Disease. PLoS Genetics, 2006, 2, e139.	3.5	82
178	Causal Associations Between Modifiable Risk Factors and the Alzheimer's Phenome. Annals of Neurology, 2021, 89, 54-65.	5.3	82
179	A genetic linkage map of the long arm of human chromosome 22. Genomics, 1989, 4, 1-6.	2.9	81
180	Novel Mutations Detected in the TSC2 Hene From Both Sporadic and Familial TSC Patients. Human Molecular Genetics, 1996, 5, 249-256.	2.9	81

#	Article	IF	CITATIONS
181	The use of animal models in the study of complex disease: all else is never equal or why do so many human studies fail to replicate animal findings?. BioEssays, 2004, 26, 170-179.	2.5	81
182	The Expanding Role of <scp>MBD</scp> Genes in Autism: Identification of a <i><scp>MECP2</scp></i> Duplication and Novel Alterations in <i><scp>MBD5</scp></i> , <i><scp>MBD6</scp></i> , and <i><scp>SETDB1</scp></i> . Autism Research, 2012, 5, 385-397.	3.8	81
183	Independent Effects of Complement Factor H Y402H Polymorphism and Cigarette Smoking on Risk of Age-Related Macular Degeneration. Ophthalmology, 2007, 114, 1151-1156.	5.2	80
184	eMERGEing progress in genomicsââ,¬â€ŧhe first seven years. Frontiers in Genetics, 2014, 5, 184.	2.3	79
185	CDKN2B-AS1 Genotype–Glaucoma Feature Correlations in Primary Open-Angle Glaucoma Patients From the United States. American Journal of Ophthalmology, 2013, 155, 342-353.e5.	3.3	76
186	Copy Number Variants in Extended Autism Spectrum Disorder Families Reveal Candidates Potentially Involved in Autism Risk. PLoS ONE, 2011, 6, e26049.	2.5	75
187	The CEPH consortium primary linkage map of human chromosome 10. Genomics, 1990, 6, 393-412.	2.9	74
188	Repeat expansions in the C9ORF72 gene contribute to Alzheimer's disease in Caucasians. Neurobiology of Aging, 2013, 34, 1519.e5-1519.e12.	3.1	74
189	<i>ABCA7</i> frameshift deletion associated with Alzheimer disease in African Americans. Neurology: Genetics, 2016, 2, e79.	1.9	74
190	Metaâ€analysis of Genomeâ€Wide Association Studies Identifies Novel Loci Associated With Optic Disc Morphology. Genetic Epidemiology, 2015, 39, 207-216.	1.3	72
191	Pitfalls of merging GWAS data: lessons learned in the eMERGE network and quality control procedures to maintain high data quality. Genetic Epidemiology, 2011, 35, 887-898.	1.3	71
192	Human $\hat{l}^2$ 2-adrenergic receptor polymorphisms: No association with essential hypertension in black or white Americans. Clinical Pharmacology and Therapeutics, 2000, 67, 670-675.	4.7	70
193	Migraine with Aura Susceptibility Locus on Chromosome 19p13 Is Distinct from the Familial Hemiplegic Migraine Locus. Genomics, 2001, 78, 150-154.	2.9	70
194	Differences in apolipoprotein E3/3 and E4/4 allele-specific gene expression in hippocampus in Alzheimer disease. Neurobiology of Disease, 2006, 21, 256-275.	4.4	70
195	ABCC9 gene polymorphism is associated with hippocampal sclerosis of aging pathology. Acta Neuropathologica, 2014, 127, 825-843.	7.7	70
196	Familial Alzheimer's disease: Progress and problems. Neurobiology of Aging, 1989, 10, 417-425.	3.1	69
197	Apolipoprotein E is associated with age at onset of amyotrophic lateral sclerosis. Neurogenetics, 2004, 5, 209-213.	1.4	69
198	Genomic convergence to identify candidate genes for Alzheimer Disease on chromosome 10. Human Mutation, 2009, 30, 463-471.	2.5	69

#	Article	IF	CITATIONS
199	<i>C9ORF72</i> Intermediate Repeat Copies Are a Significant Risk Factor for Parkinson Disease. Annals of Human Genetics, 2013, 77, 351-363.	0.8	69
200	Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. Neurobiology of Aging, 2013, 34, 357.e7-357.e19.	3.1	69
201	Two knockdown models of the autism genes SYNGAP1 and SHANK3 in zebrafish produce similar behavioral phenotypes associated with embryonic disruptions of brain morphogenesis. Human Molecular Genetics, 2015, 24, 4006-4023.	2.9	67
202	Characterization of a translocation within the von Recklinghausen neurofibromatosis region of chromosome 17. Genomics, 1989, 5, 245-249.	2.9	66
203	A Multi-Investigator/Institutional DNA Bank for AIDS-Related Human Genetic Studies: AACTG Protocol A5128. HIV Clinical Trials, 2003, 4, 287-300.	2.0	66
204	Correcting Away the Hidden Heritability. Annals of Human Genetics, 2011, 75, 348-350.	0.8	66
205	PTPRC (CD45) is not associated with the development of multiple sclerosis in U.S. patients. Nature Genetics, 2001, 29, 23-24.	21.4	65
206	Multiple susceptibility loci for multiple sclerosis. Human Molecular Genetics, 2002, 11, 2251-2256.	2.9	65
207	Enhancing linkage analysis of complex disorders: an evaluation of high-density genotyping. Human Molecular Genetics, 2004, 13, 1943-1949.	2.9	65
208	Sex differences in the genetic predictors of Alzheimer's pathology. Brain, 2019, 142, 2581-2589.	7.6	65
209	Alternative Splicing of the Tuberous Sclerosis 2 (TSC2) Gene in Human and Mouse Tissues. Genomics, 1995, 27, 475-480.	2.9	64
210	Clinical, Neuropathological and Genetic Aspects of the Tuberous Sclerosis Complex. Brain Pathology, 1995, 5, 173-179.	4.1	63
211	Endothelial Nitric Oxide Synthase Gene Variants and Primary Open-Angle Glaucoma. JAMA Ophthalmology, 2011, 129, 773.	2.4	63
212	Cross-ancestry genome-wide association analysis of corneal thickness strengthens link between complex and Mendelian eye diseases. Nature Communications, 2018, 9, 1864.	12.8	63
213	The genetic epidemiology of multiple sclerosis. Journal of Neuroimmunology, 2003, 143, 7-12.	2.3	61
214	<i>LOXL1</i> Promoter Haplotypes Are Associated with Exfoliation Syndrome in a U.S. Caucasian Population., 2011, 52, 2372.		61
215	Vitamin D receptor and Alzheimer's disease: a genetic and functional study. Neurobiology of Aging, 2012, 33, 1844.e1-1844.e9.	3.1	61
216	Vitamin D from different sources is inversely associated with Parkinson disease. Movement Disorders, 2015, 30, 560-566.	3.9	61

#	Article	IF	CITATIONS
217	Increased Prevalence of the HFE C282Y Hemochromatosis Allele in Women with Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2004, 13, 205-212.	2.5	60
218	Linkage and association with the <i>NOS2A</i> locus on chromosome 17q11 in multiple sclerosis. Annals of Neurology, 2004, 55, 793-800.	5.3	60
219	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
220	A Major Histocompatibility Class I Locus Contributes to Multiple Sclerosis Susceptibility Independently from HLA-DRB1*15:01. PLoS ONE, 2010, 5, e11296.	2.5	60
221	<pre><scp><i>PLXNA</i></scp><i>4</i> is associated with <scp>A</scp> zheimer disease and modulates tau phosphorylation. Annals of Neurology, 2014, 76, 379-392.</pre>	5.3	60
222	Prolonged Signal-Averaged P-Wave Duration as an Intermediate Phenotype for Familial Atrial Fibrillation. Journal of the American College of Cardiology, 2008, 51, 1083-1089.	2.8	59
223	Analysis of the indel at the ARMS2 3′UTR in age-related macular degeneration. Human Genetics, 2010, 127, 595-602.	3.8	59
224	Genetic Determinants of Age-Related Macular Degeneration in Diverse Populations From the PAGE Study. Investigative Ophthalmology and Visual Science, 2014, 55, 6839-6850.	3.3	59
225	Complement Factor H Increases Risk for Atrophic Age-Related Macular Degeneration. Ophthalmology, 2006, 113, 1504-1507.	5.2	58
226	Neovascular Age-Related Macular Degeneration and Its Association With LOC387715 and Complement Factor H Polymorphism. JAMA Ophthalmology, 2007, 125, 63.	2.4	58
227	Association of Rare Coding Mutations With Alzheimer Disease and Other Dementias Among Adults of European Ancestry. JAMA Network Open, 2019, 2, e191350.	5.9	58
228	Genetic Mapping of a Mouse Modifier Gene That Can Prevent ALS Onset. Genomics, 2000, 70, 181-189.	2.9	57
229	Targeted massively parallel sequencing of autism spectrum disorder-associated genes in a case control cohort reveals rare loss-of-function risk variants. Molecular Autism, 2015, 6, 43.	4.9	57
230	Huntington disease: No evidence for locus heterogeneity. Genomics, 1989, 5, 304-308.	2.9	56
231	Examination of NRCAM, LRRN3, KIAA0716, and LAMB1as autism candidate genes. BMC Medical Genetics, 2004, 5, 12.	2.1	56
232	Shared genetic contribution to ischemic stroke and Alzheimer's disease. Annals of Neurology, 2016, 79, 739-747.	5.3	56
233	Genetic Analysis of Complex Diseases. Science, 1997, 275, 1327-1330.	12.6	55
234	Design of the Genetics of Early Onset Cardiovascular Disease (GENECARD) study. American Heart Journal, 2003, 145, 602-613.	2.7	55

#	Article	IF	Citations
235	Genetic variation in the IL7RA/IL7 pathway increases multiple sclerosis susceptibility. Human Genetics, 2010, 127, 525-535.	3.8	55
236	The NEIGHBOR Consortium Primary Open-Angle Glaucoma Genome-wide Association Study. Journal of Glaucoma, 2013, 22, 517-525.	1.6	55
237	Linkage analysis for age-related macular degeneration supports a gene on chromosome 10q26. Molecular Vision, 2004, 10, 57-61.	1.1	55
238	The CALHM1 P86L Polymorphism is a Genetic Modifier of Age at Onset in Alzheimer's Disease: a Meta-Analysis Study. Journal of Alzheimer's Disease, 2010, 22, 247-255.	2.6	54
239	Increased recombination adjacent to the Huntington disease-linked D4S10 marker. Genomics, 1991, 9, 104-112.	2.9	53
240	Hemochromatosis (HFE) gene mutations and peripheral neuropathy during antiretroviral therapy. Aids, 2006, 20, 1503-1513.	2.2	53
241	Genetic variation in TP53 and risk of breast cancer in a population-based case–control study. Carcinogenesis, 2007, 28, 1680-1686.	2.8	53
242	Genetic-based prediction of disease traits: prediction is very difficult, especially about the future $\tilde{A}$ $\hat{a}$ , $\hat{A}$ . Frontiers in Genetics, 2014, 5, 162.	2.3	53
243	Early Detection of Alzheimer's Disease by Combining Apolipoprotein E and Neuroimaging. Annals of the New York Academy of Sciences, 1996, 802, 70-78.	3.8	52
244	Genome-Wide Analysis of Central Corneal Thickness in Primary Open-Angle Glaucoma Cases in the NEIGHBOR and GLAUGEN Consortia., 2012, 53, 4468.		52
245	Rare and common variants in extracellular matrix gene Fibrillin 2 (FBN2) are associated with macular degeneration. Human Molecular Genetics, 2014, 23, 5827-5837.	2.9	52
246	Association Study of Parkin Gene Polymorphisms With Idiopathic Parkinson Disease. Archives of Neurology, 2003, 60, 975.	4.5	51
247	Familial essential tremor with apparent autosomal dominant inheritance: Should we also consider other inheritance modes?. Movement Disorders, 2006, 21, 1368-1374.	3.9	51
248	Interrogating the complex role of chromosome 16p13.13 in multiple sclerosis susceptibility: independent genetic signals in the CIITA–CLEC16A–SOCS1 gene complex. Human Molecular Genetics, 2011, 20, 3517-3524.	2.9	50
249	Genetic analysis of biological pathway data through genomic randomization. Human Genetics, 2011, 129, 563-571.	3.8	50
250	Association of Genetic Variants With Primary Open-Angle Glaucoma Among Individuals With African Ancestry. JAMA - Journal of the American Medical Association, 2019, 322, 1682.	7.4	50
251	A Recurrent RNA-Splicing Mutation in the SEDL Gene Causes X-Linked Spondyloepiphyseal Dysplasia Tarda. American Journal of Human Genetics, 2001, 68, 1398-1407.	6.2	49
252	CIITA variation in the presence of HLA-DRB1*1501 increases risk for multiple sclerosis. Human Molecular Genetics, 2010, 19, 2331-2340.	2.9	49

#	Article	IF	CITATIONS
253	A genetic linkage map of chromosome 17. Genomics, 1990, 8, 1-6.	2.9	48
254	Association between bleomycin hydrolase and Alzheimer's disease in caucasians. Annals of Neurology, 1998, 44, 808-811.	5.3	48
255	Ordered subset linkage analysis supports a susceptibility locus for age-related macular degeneration on chromosome 16p12. BMC Genetics, 2004, 5, 18.	2.7	48
256	Mapping of D4S98/S114/S113 confines the Huntington's defect to a reduced physical region at the telomere of chromosome 4. Nucleic Acids Research, 1988, 16, 11769-11780.	14.5	47
257	Rare Complement Factor H Variant Associated With Age-Related Macular Degeneration in the Amish. , 2014, 55, 4455.		47
258	Advances in the genomics of common eye diseases. Human Molecular Genetics, 2013, 22, R59-R65.	2.9	46
259	Automated extraction of clinical traits of multiple sclerosis in electronic medical records. Journal of the American Medical Informatics Association: JAMIA, 2013, 20, e334-e340.	4.4	45
260	Assessing the Association of Mitochondrial Genetic Variation With Primary Open-Angle Glaucoma Using Gene-Set Analyses., 2016, 57, 5046.		44
261	A Common Variant of IL-6R is Associated with Elevated IL-6 Pathway Activity in Alzheimer's Disease Brains. Journal of Alzheimer's Disease, 2017, 56, 1037-1054.	2.6	44
262	X-linked recessive Charcot-Marie-Tooth neuropathy: Clinical and genetic study. Muscle and Nerve, 1992, 15, 368-373.	2.2	43
263	Genetic Factors in Nonsmokers with Ageâ€Related Macular Degeneration Revealed Through Genomeâ€Wide Geneâ€Environment Interaction Analysis. Annals of Human Genetics, 2013, 77, 215-231.	0.8	43
264	Identification of a Novel Locus for Febrile Seizures and Epilepsy on Chromosome 21q22. Epilepsia, 2006, 47, 1622-1628.	5.1	42
265	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
266	A Common Variant in <i>MIR182</i> Is Associated With Primary Open-Angle Glaucoma in the NEIGHBORHOOD Consortium., 2016, 57, 4528.		42
267	Global and local ancestry in Africanâ€Americans: Implications for Alzheimer's disease risk. Alzheimer's and Dementia, 2016, 12, 233-243.	0.8	42
268	Whole genome sequencing of Caribbean Hispanic families with lateâ€onset Alzheimer's disease. Annals of Clinical and Translational Neurology, 2018, 5, 406-417.	3.7	42
269	Initial Assessment of the Pathogenic Mechanisms of the Recently Identified Alzheimer Risk Loci. Annals of Human Genetics, 2013, 77, 85-105.	0.8	41
270	Rarity of the Alzheimer Disease–Protective <i>APP</i> A673T Variant in the United States. JAMA Neurology, 2015, 72, 209.	9.0	41

#	Article	IF	CITATIONS
271	Segregation of a rare $\langle i \rangle TTC3 \langle j i \rangle$ variant in an extended family with late-onset Alzheimer disease. Neurology: Genetics, 2016, 2, e41.	1.9	41
272	Early-Onset Alzheimer Disease and Candidate Risk Genes Involved in Endolysosomal Transport. JAMA Neurology, 2017, 74, 1113.	9.0	41
273	Genomeâ€Wide Association and Linkage Study in the Amish Detects a Novel Candidate Lateâ€Onset Alzheimer Disease Gene. Annals of Human Genetics, 2012, 76, 342-351.	0.8	40
274	Evidence of novel fine-scale structural variation at autism spectrum disorder candidate loci. Molecular Autism, 2012, 3, 2.	4.9	40
275	APOE Promoter Polymorphism-219T/G is an Effect Modifier of the Influence of APOE Îμ4 on Alzheimer's Disease Risk in a Multiracial Sample. Journal of Clinical Medicine, 2019, 8, 1236.	2.4	40
276	CHOROIDAL VASCULARITY INDEX AND CHOROIDAL THICKNESS IN EYES WITH RETICULAR PSEUDODRUSEN. Retina, 2020, 40, 612-617.	1.7	40
277	Using Genetic Variation and Environmental Risk Factor Data to Identify Individuals at High Risk for Age-Related Macular Degeneration. PLoS ONE, 2011, 6, e17784.	2.5	40
278	Estrogen pathway polymorphisms in relation to primary open angle glaucoma: an analysis accounting for gender from the United States. Molecular Vision, 2013, 19, 1471-81.	1.1	40
279	Joint effects of smoking history and APOE genotypes in age-related macular degeneration. Molecular Vision, 2005, 11, 941-9.	1.1	40
280	Autosomal Dominant Lateral Temporal Epilepsy: Two Families with Novel Mutations in the LGI1 Gene. Epilepsia, 2004, 45, 218-222.	5.1	39
281	A SAGE study of apolipoprotein E3/3, E3/4 and E4/4 allele-specific gene expression in hippocampus in Alzheimer disease. Molecular and Cellular Neurosciences, 2007, 36, 313-331.	2.2	39
282	Association of Long Runs of Homozygosity With Alzheimer Disease Among African American Individuals. JAMA Neurology, 2015, 72, 1313.	9.0	39
283	Discovery of gene-gene interactions across multiple independent data sets of late onset Alzheimer disease from the Alzheimer Disease Genetics Consortium. Neurobiology of Aging, 2016, 38, 141-150.	3.1	39
284	A rare missense variant of <i>CASP7</i> is associated with familial lateâ€onset Alzheimer's disease. Alzheimer's and Dementia, 2019, 15, 441-452.	0.8	39
285	Caspase-8, association with Alzheimer's Disease and functional analysis of rare variants. PLoS ONE, 2017, 12, e0185777.	2.5	38
286	Epistatic Gene-Based Interaction Analyses for Glaucoma in eMERGE and NEIGHBOR Consortium. PLoS Genetics, 2016, 12, e1006186.	3.5	38
287	Analysis of association between Alzheimer disease and the K variant of butyrylcholinesterase (BCHE-K). Neuroscience Letters, 1999, 269, 115-119.	2.1	37
288	The Carnitine Shuttle Pathway is Altered in Patients With Neovascular Age-Related Macular Degeneration., 2018, 59, 4978.		37

#	Article	IF	Citations
289	Uniparental disomy of chromosome 2 resulting in lethal trifunctional protein deficiency due to homozygous ?-subunit mutations. Human Mutation, 2002, 20, 447-451.	2.5	36
290	Lack of Association Between Autism and <i>SLC25A12</i> . American Journal of Psychiatry, 2006, 163, 929-931.	7.2	36
291	Overall Diet Quality and Age-Related Macular Degeneration. Ophthalmic Epidemiology, 2010, 17, 58-65.	1.7	36
292	Evaluating Mitochondrial DNA Variation in Autism Spectrum Disorders. Annals of Human Genetics, 2013, 77, 9-21.	0.8	36
293	Synteny on mouse chromosome 5 of homologs for human DNA loci linked to the Huntington disease gene. Genomics, 1989, 4, 419-426.	2.9	35
294	Complex gene–gene interactions in multiple sclerosis: a multifactorial approach reveals associations with inflammatory genes. Neurogenetics, 2007, 8, 11-20.	1.4	35
295	Peripheral Reticular Pigmentary Change Is Associated with Complement Factor H Polymorphism (Y402H) in Age-Related Macular Degeneration. Ophthalmology, 2008, 115, 520-524.	5.2	34
296	An association analysis of Alzheimer disease candidate genes detects an ancestral risk haplotype clade in <i>ACE</i> and putative multilocus association between <i>ACE</i> , <i>A2M</i> , and <i>LRRTM3</i> . American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 721-735.	1.7	34
297	Variation Within DNA Repair Pathway Genes and Risk of Multiple Sclerosis. American Journal of Epidemiology, 2010, 172, 217-224.	3.4	34
298	Leveraging Epidemiologic and Clinical Collections for Genomic Studies of Complex Traits. Human Heredity, 2015, 79, 137-146.	0.8	34
299	Genetic Mapping of the Batten Disease Locus (CLN3) to the Interval D16S288-D16S383 by Analysis of Haplotypes and Allelic Association. Genomics, 1994, 22, 465-468.	2.9	33
300	Linkage and association analysis of chromosome $19q13$ in multiple sclerosis. Neurogenetics, $2001$ , $3$ , $195-201$ .	1.4	33
301	Detailed Analysis of Allelic Variation in the ABCA4Gene in Age-Related Maculopathy., 2003, 44, 2868.		33
302	SNPs in Multi-Species Conserved Sequences (MCS) as useful markers in association studies: a practical approach. BMC Genomics, 2007, 8, 266.	2.8	33
303	FE65 Binds Teashirt, Inhibiting Expression of the Primate-Specific Caspase-4. PLoS ONE, 2009, 4, e5071.	2.5	33
304	Inverse Association of Female Hormone Replacement Therapy with Age-Related Macular Degeneration and Interactions with <i>ARMS2 </i> Polymorphisms., 2010, 51, 1873.		33
305	Exploring the Relationship Between Autism Spectrum Disorder and Epilepsy Using Latent Class Cluster Analysis. Journal of Autism and Developmental Disorders, 2012, 42, 1630-1641.	2.7	33
306	Analysis combining correlated glaucoma traits identifies five new risk loci for open-angle glaucoma. Scientific Reports, 2018, 8, 3124.	3.3	33

#	Article	IF	Citations
307	Alzheimer Disease: Perspectives from Epidemiology and Genetics. Journal of Law, Medicine and Ethics, 2018, 46, 694-698.	0.9	33
308	Increased <i>APOE</i> $\hat{l}\mu4$ 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
309	Analysis of the autism chromosome 2 linkage region: GAD1 and other candidate genes. Neuroscience Letters, 2004, 372, 209-214.	2.1	32
310	Phenotype Analysis of Patients With the Risk Variant LOC387715 (A69S) in Age-related Macular Degeneration. American Journal of Ophthalmology, 2008, 145, 303-307.e1.	3.3	32
311	Hypothesis-independent pathway analysis implicates GABA and Acetyl-CoA metabolism in primary open-angle glaucoma and normal-pressure glaucoma. Human Genetics, 2014, 133, 1319-1330.	3.8	32
312	Association of a Primary Open-Angle Glaucoma Genetic Risk Score With Earlier Age at Diagnosis. JAMA Ophthalmology, 2019, 137, 1190.	2.5	32
313	Analysis of Whole-Exome Sequencing Data for Alzheimer Disease Stratified by <i>APOE</i> Genotype. JAMA Neurology, 2019, 76, 1099.	9.0	32
314	Rare Variant <i>APOC3</i> R19X Is Associated With Cardio-Protective Profiles in a Diverse Population-Based Survey as Part of the Epidemiologic Architecture for Genes Linked to Environment Study. Circulation: Cardiovascular Genetics, 2014, 7, 848-853.	5.1	31
315	The Application of Genetic Risk Scores in Age-Related Macular Degeneration: A Review. Journal of Clinical Medicine, 2016, 5, 31.	2.4	31
316	Disease gene prioritization by integrating tissue-specific molecular networks using a robust multi-network model. BMC Bioinformatics, 2016, 17, 453.	2.6	31
317	Phenotypic Heterogeneity in Families with Age-related Macular Degeneration. American Journal of Ophthalmology, 1997, 124, 331-343.	3.3	30
318	Functional annotation of genomic variants in studies of late-onset Alzheimer's disease. Bioinformatics, 2018, 34, 2724-2731.	4.1	30
319	Maternal lineages and Alzheimer disease risk in the Old Order Amish. Human Genetics, 2005, 118, 115-122.	3.8	29
320	An X chromosome-wide association study in autism families identifies TBL1X as a novel autism spectrum disorder candidate gene in males. Molecular Autism, 2011, 2, 18.	4.9	29
321	Association of Smoking, Alcohol Consumption, Blood Pressure, Body Mass Index, and Glycemic Risk Factors With Age-Related Macular Degeneration. JAMA Ophthalmology, 2021, 139, 1299.	2.5	29
322	Novel Mutation in the SPG3A Gene in an African American Family With an Early Onset of Hereditary Spastic Paraplegia. Archives of Neurology, 2004, 61, 1600.	4.5	27
323	The Ubiquilin 1 Gene and Alzheimer's Disease. New England Journal of Medicine, 2005, 352, 2752-2753.	27.0	27
324	Comparing Age-related Macular Degeneration Phenotype in Probands From Singleton and Multiplex Families. American Journal of Ophthalmology, 2005, 139, 820-825.	3.3	27

#	Article	IF	CITATIONS
325	<i>CALHM1</i> Polymorphism is not Associated with Lateâ€onset Alzheimer Disease. Annals of Human Genetics, 2009, 73, 379-381.	0.8	27
326	Successful Aging Shows Linkage to Chromosomes 6, 7, and 14 in the Amish. Annals of Human Genetics, 2011, 75, 516-528.	0.8	27
327	DNA Copy Number Variants of Known Glaucoma Genes in Relation to Primary Open-Angle Glaucoma. Investigative Ophthalmology and Visual Science, 2014, 55, 8251-8258.	3.3	27
328	Rare genetic variation implicated in non-Hispanic white families with Alzheimer disease. Neurology: Genetics, 2018, 4, e286.	1.9	27
329	Genome-wide pleiotropy analysis of neuropathological traits related to Alzheimer's disease. Alzheimer's Research and Therapy, 2018, 10, 22.	6.2	27
330	Variants at chromosome 10q26 locus and the expression of HTRA1 in the retina. Experimental Eye Research, 2013, 112, 102-105.	2.6	26
331	Sex differences in the genetic architecture of cognitive resilience to Alzheimer's disease. Brain, 2022, 145, 2541-2554.	7.6	26
332	A High-Resolution Linkage Map of Human 9q34.1. Genomics, 1993, 17, 587-591.	2.9	25
333	Confronting complexity in lateâ€onset Alzheimer disease: application of twoâ€stage analysis approach addressing heterogeneity and epistasis. Genetic Epidemiology, 2008, 32, 187-203.	1.3	25
334	Resolving the relationship between ApolipoproteinE and depression. Neuroscience Letters, 2009, 455, 116-119.	2.1	25
335	The p53 Codon 72 PRO/PRO Genotype May Be Associated with Initial Central Visual Field Defects in Caucasians with Primary Open Angle Glaucoma. PLoS ONE, 2012, 7, e45613.	2.5	25
336	Linkage and association of successful aging to the 6q25 region in large Amish kindreds. Age, 2013, 35, 1467-1477.	3.0	25
337	Dissecting trait heterogeneity: a comparison of three clustering methods applied to genotypic data. BMC Bioinformatics, 2006, 7, 204.	2.6	24
338	Evidence against a role for rare ADAM10 mutations in sporadic Alzheimer Disease. Neurobiology of Aging, 2012, 33, 416-417.e3.	3.1	24
339	Linkage analyses in Caribbean Hispanic families identify novel loci associated with familial lateâ€onset Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 1397-1406.	0.8	24
340	Heritability of Choroidal Thickness in the Amish. Ophthalmology, 2016, 123, 2537-2544.	5.2	24
341	Genomeâ€wide linkage analyses of nonâ€Hispanic white families identify novel loci for familial lateâ€onset Alzheimer's disease. Alzheimer's and Dementia, 2016, 12, 2-10.	0.8	24
342	NR1H3 p.Arg415Gln Is Not Associated to Multiple Sclerosis Risk. Neuron, 2016, 92, 333-335.	8.1	24

#	Article	IF	CITATIONS
343	Male-specific epistasis between WWC1 and TLN2 genes is associated with Alzheimer's disease. Neurobiology of Aging, 2018, 72, 188.e3-188.e12.	3.1	24
344	Refined mapping and characterization of the recessive familial amyotrophic lateral sclerosis locus () Tj ETQq0 0 0 $^{\circ}$	rgBŢ /Ove	rlock 10 Tf 5
345	Investigation of seven proposed regions of linkage in multiple sclerosis: an American and French collaborative study. Neurogenetics, 2004, 5, 45-48.	1.4	23
346	Lack of support for association between the KIF1B rs10492972[C] variant and multiple sclerosis. Nature Genetics, 2010, 42, 469-470.	21.4	23
347	Population Differences in Genetic Risk for Age-Related Macular Degeneration and Implications for Genetic Testing. JAMA Ophthalmology, 2012, 130, 116.	2.4	23
348	Mitochondrial Haplogroup X is associated with successful aging in the Amish. Human Genetics, 2012, 131, 201-208.	3.8	23
349	Enhancing the Power of Genetic Association Studies through the Use of Silver Standard Cases Derived from Electronic Medical Records. PLoS ONE, 2013, 8, e63481.	2.5	23
350	Association of <i>APOE</i> With Primary Open-Angle Glaucoma Suggests a Protective Effect for <i>APOE <math>\hat{l}\mu</math>4</i> . , 2020, 61, 3.		23
351	Comprehensive association analysis of APOE regulatory region polymorphisms in Alzheimer disease. Neurogenetics, 2004, 5, 201-208.	1.4	22
352	Haplotypes Spanning the Complement Factor H Gene Are Protective against Age-Related Macular Degeneration., 2007, 48, 4277.		22
353	THE ARMS2 A69S VARIANT AND BILATERAL ADVANCED AGE-RELATED MACULAR DEGENERATION. Retina, 2012, 32, 1486-1491.	1.7	22
354	Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimer's Disease Sequencing Project. Dementia and Geriatric Cognitive Disorders, 2018, 45, 1-17.	1.5	22
355	One for all and all for One: Improving replication of genetic studies through network diffusion. PLoS Genetics, 2018, 14, e1007306.	3.5	22
356	Chromlook: An interactive program for error detection and mapping in reference linkage data. Genomics, 1992, 14, 517-519.	2.9	21
357	Linkage analysis of candidate myelin genes in familial multiple sclerosis. Neurogenetics, 1999, 2, 155-162.	1.4	21
358	Joint Analysis of Nuclear and Mitochondrial Variants in Age-Related Macular Degeneration Identifies Novel Loci <i>TRPM1</i> and <i>ABHD2/RLBP1</i> , 2017, 58, 4027.		21
359	Lack of association of polymorphisms in homocysteine metabolism genes with pseudoexfoliation syndrome and glaucoma. Molecular Vision, 2008, 14, 2484-91.	1.1	21
360	A genetic linkage map of human chromosome 9q. Genomics, 1992, 14, 715-720.	2.9	20

#	Article	IF	CITATIONS
361	Genetic and physical mapping of the GLUR5 glutamate receptor gene on human chromosome 21. Human Genetics, 1994, 94, 565-570.	3.8	20
362	Enabling Genomic-Phenomic Association Discovery without Sacrificing Anonymity. PLoS ONE, 2013, 8, e53875.	2.5	20
363	Biologyâ€Driven Geneâ€Gene Interaction Analysis of Ageâ€Related Cataract in the eMERGE Network. Genetic Epidemiology, 2015, 39, 376-384.	1.3	20
364	No association between the HLA-A2 allele and Alzheimer disease. Neurogenetics, 1999, 2, 177-182.	1.4	19
365	PCDH11X variation is not associated with late-onset Alzheimer disease susceptibility. Psychiatric Genetics, 2010, 20, 321-324.	1.1	19
366	Evidence for CRHR1 in multiple sclerosis using supervised machine learning and meta-analysis in 12 566 individuals. Human Molecular Genetics, 2010, 19, 4286-4295.	2.9	19
367	Identification and Confirmation of an Exonic Splicing Enhancer Variation in Exon 5 of the Alzheimer Disease Associated <i>PICALM</i> Gene. Annals of Human Genetics, 2012, 76, 448-453.	0.8	19
368	Relationship Between Depressive Symptoms and Cognition in Older, Non-demented African Americans. Journal of the International Neuropsychological Society, 2014, 20, 756-763.	1.8	19
369	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
370	Genomeâ€wide association and multiâ€omics studies identify <i>MGMT</i> as a novel risk gene for Alzheimer's disease among women. Alzheimer's and Dementia, 2023, 19, 896-908.	0.8	19
371	A locus at 19q13.31 significantly reduces the ApoE Îμ4 risk for Alzheimer's Disease in African Ancestry. PLoS Genetics, 2022, 18, e1009977.	3.5	19
372	Lack of association between autism and four heavy metal regulatory genes. NeuroToxicology, 2011, 32, 769-775.	3.0	18
373	Progression Rate From Intermediate to Advanced Age-Related Macular Degeneration Is Correlated With the Number of Risk Alleles at the CFH Locus. , 2016, 57, 6107.		18
374	Genetic correlations between intraocular pressure, blood pressure and primary open-angle glaucoma: a multi-cohort analysis. European Journal of Human Genetics, 2017, 25, 1261-1267.	2.8	18
375	Protein phosphatase 2A and complement component 4 are linked to the protective effect of <i>APOE</i> £,2 for Alzheimer's disease. Alzheimer's and Dementia, 2022, 18, 2042-2054.	0.8	18
376	The GABBR1 locus and the G1465A variant is not associated with temporal lobe epilepsy preceded by febrile seizures. BMC Medical Genetics, 2005, 6, 13.	2.1	17
377	AMISH EYE STUDY. Retina, 2019, 39, 1540-1550.	1.7	17
378	Degree of heteroplasmy reflects oxidant damage in a large family with the mitochondrial DNA A8344G mutation. Free Radical Biology and Medicine, 2005, 38, 678-683.	2.9	16

#	Article	IF	Citations
379	A prevalenceâ€based association test for caseâ€control studies. Genetic Epidemiology, 2008, 32, 600-605.	1.3	16
380	Analysis of Single Nucleotide Polymorphisms in the <i>NOS2A</i> Gene and Interaction with Smoking in Ageâ€Related Macular Degeneration. Annals of Human Genetics, 2010, 74, 195-201.	0.8	16
381	The Impact of the Human Genome Project on Complex Disease. Genes, 2014, 5, 518-535.	2.4	16
382	A population-specific reference panel empowers genetic studies of Anabaptist populations. Scientific Reports, 2017, 7, 6079.	3.3	16
383	Association of Rare <i>CYP39A1</i> Variants With Exfoliation Syndrome Involving the Anterior Chamber of the Eye. JAMA - Journal of the American Medical Association, 2021, 325, 753.	7.4	16
384	Automated identification of clinical features from sparsely annotated 3-dimensional medical imaging. Npj Digital Medicine, 2021, 4, 44.	10.9	16
385	Using the PhenX Toolkit to Select Standard Measurement Protocols for Your Research Study. Current Protocols, 2021, 1, e149.	2.9	16
386	Bringing the genetics of macular degeneration into focus. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 16725-16726.	7.1	15
387	Effect of heterogeneity on the chromosome 10 risk in late-onset Alzheimer disease. Human Mutation, 2007, 28, 1065-1073.	2.5	15
388	Genome-Wide Association Studies: Getting to Pathogenesis, the Role of Inflammation/Complement in Age-Related Macular Degeneration. Cold Spring Harbor Perspectives in Medicine, 2014, 4, a017186-a017186.	6.2	15
389	Association of clusterin (CLU) variants and exfoliation syndrome: An analysis in two Caucasian studies and a meta-analysis. Experimental Eye Research, 2015, 139, 115-122.	2.6	15
390	The Relationship Between Reticular Pseudodrusen and Severity of AMD. Ophthalmology, 2016, 123, 921-923.	5.2	15
391	Polygenic Risk Score for Alzheimer's Disease in Caribbean Hispanics. Annals of Neurology, 2021, 90, 366-376.	5.3	15
392	Mutations in GABRA1, GABRA5, GABRG2 and GABRD receptor genes are not a major factor in the pathogenesis of familial focal epilepsy preceded by febrile seizures. Neuroscience Letters, 2006, 394, 74-78.	2.1	14
393	Genotype at Polymorphism rs11200638 and HTRA1 Expression Level. JAMA Ophthalmology, 2010, 128, 1491.	2.4	14
394	Testosterone Pathway Genetic Polymorphisms in Relation to Primary Open-Angle Glaucoma: An Analysis in Two Large Datasets., 2018, 59, 629.		14
395	Education Moderates the Relation Between APOE É>4 and Memory in Nondemented Non-Hispanic Black Older Adults. Journal of Alzheimer's Disease, 2019, 72, 495-506.	2.6	14
396	Reduced plasma haptoglobin and urinary taurine in familial seizures identified through the multisib strategy. American Journal of Medical Genetics Part A, 1986, 24, 723-734.	2.4	13

#	Article	IF	CITATIONS
397	PRNP M129V homozygosity in multiple system atrophy vs. Parkinson's disease. Clinical Autonomic Research, 2008, 18, 13-19.	2.5	13
398	<i>APOE</i> is not Associated with Alzheimer Disease: a Cautionary tale of Genotype Imputation. Annals of Human Genetics, 2010, 74, 189-194.	0.8	13
399	Reproducibility of qualitative assessment of drusen volume in eyes with age related macular degeneration. Eye, 2021, 35, 2594-2600.	2.1	13
400	Examination of Candidate Exonic Variants for Association to Alzheimer Disease in the Amish. PLoS ONE, 2015, 10, e0118043.	2.5	13
401	Plasma Metabolomics of Intermediate and Neovascular Age-Related Macular Degeneration Patients. Cells, 2021, 10, 3141.	4.1	13
402	No Association between ?1-Antichymotrypsin and Familial Alzheimer's Diseases. Annals of the New York Academy of Sciences, 1996, 802, 35-41.	3.8	12
403	The Q7R Saitohin gene polymorphism is not associated with Alzheimer disease. Neuroscience Letters, 2003, 347, 143-146.	2.1	12
404	Genetic Association Analysis of Drusen Progression. , 2016, 57, 2225.		12
405	Genetic Correlations Between Diabetes and Glaucoma: An Analysis of Continuous and Dichotomous Phenotypes. American Journal of Ophthalmology, 2019, 206, 245-255.	3.3	12
406	Use of local genetic ancestry to assess <i>TOMM40</i> -523′ and risk for Alzheimer disease. Neurology: Genetics, 2020, 6, e404.	1.9	12
407	Whole exome sequencing of extreme age-related macular degeneration phenotypes. Molecular Vision, 2016, 22, 1062-76.	1.1	12
408	Progranulin mutations in clinical and neuropathological Alzheimer's disease. Alzheimer's and Dementia, 2022, 18, 2458-2467.	0.8	12
409	Admixture Mapping of Alzheimer's disease in Caribbean Hispanics identifies a new locus on 22q13.1. Molecular Psychiatry, 2022, 27, 2813-2820.	7.9	12
410	Manifestations of Alzheimer's disease genetic risk in the blood are evident in a multiomic analysis in healthy adults aged 18 to 90. Scientific Reports, 2022, 12, 6117.	3.3	12
411	Combinatorial Mismatch Scan (CMS) for loci associated with dementia in the Amish. BMC Medical Genetics, 2006, 7, 19.	2.1	11
412	Determining Genetic Component of a Disease., 0,, 91-115.		11
413	Methods for analysis in pharmacogenomics: lessons from the Pharmacogenetics Research Network Analysis Group. Pharmacogenomics, 2009, 10, 243-251.	1.3	11
414	A novel ARMS2 splice variant is identified in human retina. Experimental Eye Research, 2012, 94, 187-191.	2.6	11

#	Article	IF	CITATIONS
415	DNA variants in <i>CACNA1C</i> modify Parkinson disease risk only when vitamin D level is deficient. Neurology: Genetics, 2016, 2, e72.	1.9	11
416	Family-Based Genome-Wide Association Study of South Indian Pedigrees Supports < i>WNT7B < /i> as a Central Corneal Thickness Locus., 2018, 59, 2495.		11
417	Extracting Primary Open-Angle Glaucoma from Electronic Medical Records for Genetic Association Studies. PLoS ONE, 2015, 10, e0127817.	2.5	11
418	Partial linkage map of chromosome 13q in the region of the Wilson disease and retinoblastoma genes. Genetic Epidemiology, 1988, 5, 375-380.	1.3	10
419	A genetic map of chromosome 1: Comparison of different data sets and linkage programs. Genomics, 1990, 7, 313-318.	2.9	10
420	Linkage analysis in juvenile neuronal ceroid lipofuscinosis. American Journal of Medical Genetics Part A, 1992, 42, 542-545.	2.4	10
421	Charcot-marie-tooth neuropathy related to chromosome 1. American Journal of Medical Genetics Part A, 1992, 42, 728-732.	2.4	10
422	A genetic linkage map of the chromosome 4 short arm. Somatic Cell and Molecular Genetics, 1993, 19, 95-101.	0.7	10
423	Consortium Fine Localization of X-Linked Charcot-Marie-Tooth Disease (CMTX1): Additional Support that Connexin32 Is the Defect in CMTX1. Human Heredity, 1995, 45, 121-128.	0.8	10
424	Association of <i>CYP1B1</i> Haplotypes and Breast Cancer Risk in Caucasian Women. Cancer Epidemiology Biomarkers and Prevention, 2009, 18, 1321-1323.	2.5	10
425	Hamilton et al. Respond to "Consolidating Data Harmonization". American Journal of Epidemiology, 2011, 174, 265-266.	3.4	10
426	Dissection of Chromosome 16p12 Linkage Peak Suggests a Possible Role for CACNG3 Variants in Age-Related Macular Degeneration Susceptibility., 2011, 52, 1748.		10
427	Pathway Analysis Integrating Genome-Wide and Functional Data Identifies <i>PLCG2</i> as a Candidate Gene for Age-Related Macular Degeneration., 2019, 60, 4041.		10
428	A comparative analysis of the information content in long and short SAGE libraries. BMC Bioinformatics, 2006, 7, 504.	2.6	9
429	Overview of Linkage Analysis in Complex Traits. Current Protocols in Human Genetics, 2010, 64, Unit 1.9.1-18.	3.5	9
430	A Genomeâ€Wide Linkage Screen in the Amish with Parkinson Disease Points to Chromosome 6. Annals of Human Genetics, 2011, 75, 351-358.	0.8	9
431	Estimating cumulative pathway effects on risk for age-related macular degeneration using mixed linear models. BMC Bioinformatics, 2015, 16, 329.	2.6	9
432	Pathway analysis by randomization incorporating structureâ€"PARIS: an update. Bioinformatics, 2016, 32, 2361-2363.	4.1	9

#	Article	IF	Citations
433	Quality Control for the Illumina HumanExome BeadChip. Current Protocols in Human Genetics, 2016, 90, 2.14.1-2.14.16.	3.5	9
434	Assay for Transposaseâ€Accessible Chromatin Using Sequencing (ATACâ€seq) Data Analysis. Current Protocols in Human Genetics, 2017, 92, 20.4.1-20.4.13.	3.5	9
435	Analysis of brain region-specific co-expression networks reveals clustering of established and novel genes associated with Alzheimer disease. Alzheimer's Research and Therapy, 2020, 12, 103.	6.2	9
436	Association of mitochondrial variants and haplogroups identified by whole exome sequencing with Alzheimer's disease. Alzheimer's and Dementia, $2021$ , , .	0.8	9
437	KIAA1462, A Coronary Artery Disease Associated Gene, Is a Candidate Gene for Late Onset Alzheimer Disease in APOE Carriers. PLoS ONE, 2013, 8, e82194.	2.5	9
438	The National Institute on Aging Lateâ€Onset Alzheimer's Disease Family Based Study: A resource for genetic discovery. Alzheimer's and Dementia, 2022, 18, 1889-1897.	0.8	9
439	Feasibility of High-Throughput Genome-Wide Genotyping using DNA from Stored Buccal Cell Samples. Biomarker Insights, 2010, 5, BMI.S5062.	2.5	8
440	Coding Variants in ARMS2 and the Risk of Age-Related Macular Degeneration. JAMA Ophthalmology, 2013, 131, 804.	2.5	8
441	The intelligent use and clinical benefits of electronic medical records in multiple sclerosis. Expert Review of Clinical Immunology, 2015, 11, 205-211.	3.0	8
442	CpGâ€related SNPs in the MS4A region have a doseâ€dependent effect on risk of late–onset Alzheimer disease. Aging Cell, 2019, 18, e12964.	6.7	8
443	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
444	Parsing the genetic heterogeneity of chromosome 12q susceptibility genes for Alzheimer disease by family-based association analysis. Neurogenetics, 2006, 7, 157-165.	1.4	7
445	Putting Pleiotropy and Selection Into Context Defines a New Paradigm for Interpreting Genetic Data. Circulation: Cardiovascular Genetics, 2013, 6, 299-307.	5.1	7
446	Rare variants and loci for age-related macular degeneration in the Ohio and Indiana Amish. Human Genetics, 2019, 138, 1171-1182.	3.8	7
447	Evaluating Power and Type 1 Error in Large Pedigree Analyses of Binary Traits. PLoS ONE, 2013, 8, e62615.	2.5	7
448	Peripherin gene is linked to keratin 18 gene on human chromosome 12. Somatic Cell and Molecular Genetics, 1995, 21, 83-88.	0.7	6
449	Microduplications in an autism multiplex family narrow the region of susceptibility for developmental disorders on 15q24 and implicate 7p21. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 493-501.	1.7	6
450	Age at natural menopause genetic risk score in relation to age at natural menopause and primary open-angle glaucoma in a US-based sample. Menopause, 2017, 24, 150-156.	2.0	6

#	Article	IF	CITATIONS
451	Immune and Inflammatory Pathways Implicated by Whole Blood Transcriptomic Analysis in a Diverse Ancestry Alzheimer's Disease Cohort. Journal of Alzheimer's Disease, 2020, 76, 1047-1060.	2.6	6
452	Automating Data Manipulation for Genetic Analysis Using a Data Base Management System. Human Heredity, 1985, 35, 296-301.	0.8	5
453	Association Analysis of Genetic Polymorphisms in the CDC2 Gene with Late-Onset Alzheimer Disease. Dementia and Geriatric Cognitive Disorders, 2007, 23, 126-132.	1.5	5
454	Rapid Dissection of the Genetic Risk of Age-Related Macular Degeneration. JAMA - Journal of the American Medical Association, 2007, 297, 401.	7.4	5
455	No Association between SNP rs498055 on Chromosome 10 and Late-Onset Alzheimer Disease in Multiple Datasets. Annals of Human Genetics, 2007, 72, 070824035807001-???.	0.8	5
456	Beyond proof of principle: new genes for Alzheimer's disease through collaboration. Lancet Neurology, The, 2009, 8, 977-979.	10.2	5
457	Lack of Association of Polymorphisms in Elastin With Pseudoexfoliation Syndrome and Glaucoma. Journal of Glaucoma, 2010, 19, 432-436.	1.6	5
458	A new locus for autosomal dominant generalized epilepsy associated with mild mental retardation on chromosome 3p. Epilepsia, 2011, 52, 993-999.	5.1	5
459	Size matters: How population size influences genotype–phenotype association studies in anonymized data. Journal of Biomedical Informatics, 2014, 52, 243-250.	4.3	5
460	Set-Based Joint Test of Interaction Between SNPs in the VEGF Pathway and Exogenous Estrogen Finds Association With Age-Related Macular Degeneration. , 2014, 55, 4873.		5
461	The molecular genetics of eye diseases. Human Molecular Genetics, 2017, 26, R1-R1.	2.9	5
462	Identification of rare noncoding sequence variants in gamma-aminobutyric acid A receptor, alpha 4 subunit in autism spectrum disorder. Neurogenetics, 2018, 19, 17-26.	1.4	5
463	COMPARISON OF SPECTRALIS AND CIRRUS OPTICAL COHERENCE TOMOGRAPHY FOR THE DETECTION OF INCOMPLETE AND COMPLETE RETINAL PIGMENT EPITHELIUM AND OUTER RETINAL ATROPHY. Retina, 2021, 41, 1851-1857.	1.7	5
464	An association test of the spatial distribution of rare missense variants within protein structures identifies Alzheimer's disease–related patterns. Genome Research, 2022, 32, 778-790.	5.5	5
465	PowerTrim: An Automated Decision Support Algorithm for Preprocessing Family-Based Genetic Data. American Journal of Human Genetics, 2003, 72, 1280-1281.	6.2	4
466	Whole-Genome Amplification of Oral Rinse Self-Collected DNA in a Population-Based Case-Control Study of Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 1610-1614.	2.5	4
467	The importance of rare DNA variation in neurologic disease. Neurology, 2013, 80, 974-975.	1.1	4
468	[O1–03–01]: GENOMEâ€WIDE RARE VARIANT IMPUTATION AND TISSUEâ€SPECIFIC TRANSCRIPTOMIC ANAI IDENTIFY NOVEL RARE VARIANT CANDIDATE LOCI IN LATEâ€ONSET ALZHEIMER's DISEASE: THE ALZHEIMER's DISEASE GENETICS CONSORTIUM. Alzheimer's and Dementia, 2017, 13, P189.	LYSIS 0.8	4

#	Article	IF	CITATIONS
469	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
470	Largeâ€scale sequencing studies expand the known genetic architecture of Alzheimer's disease. Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring, 2021, 13, e12255.	2.4	4
471	Genetic variants in the <i>SHISA6</i> gene are associated with delayed cognitive impairment in two family datasets. Alzheimer's and Dementia, 2023, 19, 611-620.	0.8	4
472	Nonallelic Heterogeneity in Autosomal Dominant Retinitis Pigmentosa with Incomplete Penetrance. Genomics, 1994, 22, 659-660.	2.9	3
473	Examination of candidate genes in language disorder: A model of genetic association for treatment studies. Mental Retardation and Developmental Disabilities Research Reviews, 2004, 10, 208-217.	3.6	3
474	Defining Disease Phenotypes., 0,, 51-89.		3
475	Leveraging Identity-by-Descent for Accurate Genotype Inference in Family Sequencing Data. PLoS Genetics, 2015, 11, e1005271.	3.5	3
476	Pedigree Selection and Information Content. Current Protocols in Human Genetics, 2018, 97, e56.	3.5	3
477	Effect of OCT B-Scan Density on Sensitivity for Detection of Intraretinal Hyperreflective Foci in Eyes with Age-Related Macular Degeneration. Current Eye Research, 2022, 47, 1294-1299.	1.5	3
478	The glycinamide ribonucleotide transformylase (GART) gene is not responsible for familial amyotrophic lateral sclerosis. Neuromuscular Disorders, 1993, 3, 157-160.	0.6	2
479	No association of $\hat{l}\pm 1$ -antichymotrypsin flanking region polymorphism and Alzheimer disease risk in early- and late-onset Alzheimer disease patients. Neuroscience Letters, 1998, 250, 79-82.	2.1	2
480	O1-03-03: Identification of Novel Candidate Genes for Early-Onset Alzheimer's Disease Through Integrated Whole-Exome Sequencing and Exome Chip Array Association Analysis., 2016, 12, P177-P178.		2
481	Introducing COCOS: codon consequence scanner for annotating reading frame changes induced by stop-lost and frame shift variants. Bioinformatics, 2017, 33, btw820.	4.1	2
482	Genotype Correlation Analysis Reveals Pathway-Based Functional Disequilibrium and Potential Epistasis in the Human Interactome. Lecture Notes in Computer Science, 2014, 8602, 890-901.	1.3	2
483	DRUG-DRUG INTERACTION PROFILES OF MEDICATION REGIMENS EXTRACTED FROM A DE-IDENTIFIED ELECTRONIC MEDICAL RECORDS SYSTEM. AMIA Summits on Translational Science Proceedings, 2016, 2016, 33-40.	0.4	2
484	Genome-wide association study of brain arteriolosclerosis. Journal of Cerebral Blood Flow and Metabolism, 2022, 42, 1437-1450.	4.3	2
485	Genetic architecture of RNA editing regulation in Alzheimer's disease across diverse ancestral populations. Human Molecular Genetics, 2022, 31, 2876-2886.	2.9	2
486	APOEâ€stratified genomeâ€wide association analysis identifies novel Alzheimer disease candidate risk loci for African Americans. Alzheimer's and Dementia, 2021, 17, e056383.	0.8	2

#	Article	IF	CITATIONS
487	Artificial intelligence-based strategies to identify patient populations and advance analysis in age-related macular degeneration clinical trials. Experimental Eye Research, 2022, 220, 109092.	2.6	2
488	Evaluation of screening strategies to detect an oligogenic disease. Genetic Epidemiology, 1995, 12, 665-669.	1.3	1
489	Linkage of Parkinsonism and Alzheimer's disease with Lewy body pathology to chromosome 12. Annals of Neurology, 2002, 52, 524-524.	5.3	1
490	[O2–08–03]: WHOLEâ€GENOME SEQUENCING IN FAMILIAL LATEâ€ONSET ALZHEIMER's DISEASE IDENTIFIES VARIATION IN AD CANDIDATE GENES. Alzheimer's and Dementia, 2017, 13, P571.	RARE 0.8	1
491	Using linkage analysis to identify novel geneâ€gene interactions in Alzheimer's disease. Alzheimer's and Dementia, 2020, 16, e043435.	0.8	1
492	Genomeâ€wide metaâ€analysis of lateâ€onset Alzheimer's disease using rare variant imputation in 65,602 subjects identifies risk loci with roles in memory, neurodevelopment, and cardiometabolic traits: The international genomics of Alzheimer's project (IGAP). Alzheimer's and Dementia, 2020, 16, e044193.	0.8	1
493	iPSCâ€derived neurons and microglia with an Africanâ€specific ABCA7 frameshift deletion have impaired function. Alzheimer's and Dementia, 2020, 16, e046109.	0.8	1
494	Exome sequencing identifies rare damaging variants in the ATB8B4 and ABCA1 genes as novel risk factors for Alzheimer's disease Alzheimer's and Dementia, 2021, 17 Suppl 3, e055982.	0.8	1
495	The genetic architecture of Alzheimer disease risk in the Ohio and Indiana Amish. Human Genetics and Genomics Advances, 2022, 3, 100114.	1.7	1
496	Analysis of Huntington disease linkage to G8. Genetic Epidemiology, 1986, 3, 223-228.	1.3	0
497	Genetic Linkage Reference Maps: Access to Internetâ€Based Resources. Current Protocols in Human Genetics, 1998, 18, Appendix 5.	3.5	O
498	Hunting genetic diseases: Exploring a multistage approach to identifying disease loci. Genetic Epidemiology, 1999, 17, S557-S562.	1.3	0
499	THE GENETICS AND EPIDEMIOLOGY OF MULTIPLE SCLEROSIS., 2003,,.		O
500	Complex Genetic Interactions. , 0, , 397-421.		0
501	Genomics and Bioinformatics., 0,, 423-454.		O
502	Designing a Study for Identifying Genes in Complex Traits., 0,, 455-468.		0
503	Data Analysis Issues in Expression Profiling. , 0, , 193-217.		O
504	Quantitative Trait Linkage Analysis., 0,, 237-253.		0

#	Article	IF	CITATIONS
505	Response to Letter Regarding Article, "Cardiac Sodium Channel ( SCN5A ) Variants Associated with Atrial Fibrillation― Circulation, 2008, 118, .	1.6	0
506	A Primer in Statistical Methods in Genetics. , 0, , 20-32.		0
507	PL-01-01: Genetics of Alzheimer's disease: Progress and future. , 2010, 6, S60-S60.		O
508	O5â€03â€01: Deep resequencing of 9 confirmed lateâ€onset Alzheimer's disease (LOAD) loci identifies multiple genomic regions with potentially functional variants. Alzheimer's and Dementia, 2012, 8, P734.	0.8	0
509	Genetics of Alzheimer Disease. , 2013, , 1-20.		O
510	P3â€082: Assessment of the Genetic Variance of Lateâ€Onset Alzheimer's Disease. Alzheimer's and Dementia, 2016, 12, P849.	'0.8	0
511	O1â€03â€02: <i>ABCA7</i> Frameshift Deletion Associated with Alzheimer's Disease in African Americans. Alzheimer's and Dementia, 2016, 12, P177.	0.8	O
512	O1-03-05: High-Resolution Imputation in Genome-Wide Association Studies of Late-Onset Alzheimer's Disease Identifies Novel Rare Variant Associations. , 2016, 12, P178-P179.		0
513	O1-09-02: Whole Exome Sequencing of Late Onset Multiplex Families Identifies Rare Coding Variants in Known and Novel Alzheimer's Disease Genes. , 2016, 12, P196-P197.		O
514	O1â€09â€03: Whole Genome Sequencing in Familial Lateâ€Onset Alzheimer's Disease Identifies Variations in TTC3 and FSIP2. Alzheimer's and Dementia, 2016, 12, P197.	0.8	0
515	P1â€122: Multivariate Phenotypes Association Study of Neuropathological Features of Alzheimer's Disease and Related Dementias. Alzheimer's and Dementia, 2016, 12, P450.	0.8	O
516	[O2–08–02]: SEX‧PECIFIC ANALYSIS OF THE ADSP CASE ONTROL WHOLE XOME SEQUENCING DATAlzheimer's and Dementia, 2017, 13, P571.	TASET.	0
517	O3â€13â€01: HIGHLY PENETRANT LATEâ€ONSET ALZHEIMER DISEASE VARIANTS IN NOTCH3 IN ASHKENAZI JEWS Alzheimer's and Dementia, 2019, 15, P918.	·0.8	0
518	Segregation, linkage, GWAS, and sequencing., 2020, , 7-23.		0
519	Statistical driver genes as a means to uncover missing heritability for age-related macular degeneration. BMC Medical Genomics, 2020, 13, 95.	1.5	0
520	Comparative transâ€ethnic metaâ€analysis of whole exome sequencing variation for Alzheimer's disease (AD) in 18,402 individuals of the Alzheimer's Disease Sequencing Project (ADSP). Alzheimer's and Dementia, 2020, 16, e041583.	0.8	0
521	Sex differences in genetic predictors of resilience to Alzheimer's disease. Alzheimer's and Dementia, 2020, 16, e043259.	0.8	O
522	Longitudinal assessment of cognitive decline in the Amish. Alzheimer's and Dementia, 2020, 16, e043440.	0.8	0

#	Article	IF	CITATIONS
523	Mechanism for the protective effect of APOE $\hat{l}\mu 2$ against Alzheimer disease is linked to tau and the classical complement pathway. Alzheimer's and Dementia, 2020, 16, e044881.	0.8	O
524	Search for protective genetic variants in Alzheimer disease in the U.S. Midwestern Amish. Alzheimer's and Dementia, 2020, 16, e045350.	0.8	0
525	A multiancestry analysis of Alzheimer's disease coexpressed gene networks identifies a common immune signaling pathway regulated by granulocyteâ€colony stimulating factor (Gâ€CSF). Alzheimer's and Dementia, 2020, 16, e045361.	0.8	0
526	Increased <i>APOEâ€e4</i> expression is associated with reactive A1 astrocytes and may confer the difference in Alzheimer disease risk from different ancestral backgrounds. Alzheimer's and Dementia, 2020, 16, e045415.	0.8	0
527	Assessing whole genome sequencing variation for Alzheimer's disease in 4707 individuals from the Alzheimer's Disease Sequencing Project (ADSP). Alzheimer's and Dementia, 2020, 16, e045548.	0.8	0
528	Transcriptomic characterization of a Puerto Rican Alzheimer disease cohort implicates convergent immuneâ€related pathways. Alzheimer's and Dementia, 2020, 16, e045890.	0.8	0
529	Mapping Alzheimer disease–associated regions in the African American population. Alzheimer's and Dementia, 2020, 16, e046072.	0.8	0
530	Education and its effect on risk and age at onset in Alzheimer disease (AD) in African Americans. Alzheimer's and Dementia, 2020, 16, e046078.	0.8	0
531	Genomeâ€wide interaction study of smoking in Alzheimer's disease. Alzheimer's and Dementia, 2020, 16, e046149.	0.8	0
532	Structural characterization of rare missense variants within known neurodegenerative disease proteins. Alzheimer's and Dementia, 2020, 16, e046405.	0.8	0
533	Joint linkage and association mapping of preserved cognition in the oldâ€order Amish. Alzheimer's and Dementia, 2020, 16, e046416.	0.8	0
534	The GGLEAM Study: Understanding Glaucoma in the Ohio Amish. International Journal of Environmental Research and Public Health, 2021, 18, 1551.	2.6	0
535	Hadoop and PySpark for reproducibility and scalability of genomic sequencing studies. Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing, 2020, 25, 523-534.	0.7	0
536	Assessment of ADâ€related plasma biomarkers in diverse ancestral populations. Alzheimer's and Dementia, 2021, 17, .	0.8	0
537	Does higher educational attainment influence functional capabilities among African Americans with Alzheimer $\hat{a} \in \mathbb{R}^{M}$ s disease?. Alzheimer's and Dementia, 2021, 17, .	0.8	0
538	Association of a locus on chromosome 17 with earlier age at onset of cognitive impairment in a familial Amish dataset. Alzheimer's and Dementia, 2021, 17, e056288.	0.8	0
539	Genomeâ€wide association for protective variants in Alzheimer's disease in the Midwestern Amish. Alzheimer's and Dementia, 2021, 17, e056363.	0.8	0
540	Preferential preservation of constructional praxis delayed recall compared to word list delayed recall in the Amish. Alzheimer's and Dementia, 2021, 17, e056386.	0.8	0

#	Article	IF	CITATIONS
541	Genetic risk score for Alzheimer's disease in the Amish highlights differences in the genetic architecture compared to other European ancestry populations Alzheimer's and Dementia, 2021, 17 Suppl 3, e053304.	0.8	0
542	Genome-wide association and multi-omics studies identify MGMT as a novel risk gene for Alzheimer disease among women Alzheimer's and Dementia, 2021, 17 Suppl 3, e054483.	0.8	0
543	Multiple viruses detected in human DNA are associated with Alzheimer disease risk Alzheimer's and Dementia, 2021, 17 Suppl 3, e054585.	0.8	0
544	Sex differences in the genetic architecture underlying resilience in AD Alzheimer's and Dementia, 2021, 17 Suppl 3, e055010.	0.8	0
545	Sex-specific genetic predictors of memory performance Alzheimer's and Dementia, 2021, 17 Suppl 3, e056083.	0.8	0
546	Expression quantitative trait loci (eQTL) analysis in a diverse Alzheimer disease cohort reveals ancestry-specific regulatory architectures Alzheimer's and Dementia, 2021, 17 Suppl 3, e056211.	0.8	0
547	Suggestive linkage and association of preserved cognition to chromosome 18 in genetically at-risk Amish Alzheimer's and Dementia, 2021, 17 Suppl 3, e056306.	0.8	0
548	Admixture mapping identifies novel regions influencing Alzheimer disease in African Americans Alzheimer's and Dementia, 2021, 17 Suppl 3, e056443.	0.8	0
549	Genome-wide association study of cognitive status and decline in the Amish Alzheimer's and Dementia, 2021, 17 Suppl 3, e056525.	0.8	0