Jonathan L Haines

List of Publications by Citations

550 papers	64,609 citations	112 h-index	246 g-index
612 ext. papers	75,011 ext. citations	9.6 avg, IF	6.4 L-index

#	Paper	IF	Citations
550	Finding the missing heritability of complex diseases. *Nature*, **2009**, 461, 747-53	50.4	6084
549	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. *Nature*, **1993**, 362, 59-62	50.4	5355
548	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
547	Complement factor H variant increases the risk of age-related macular degeneration. *Science*, **2005**, 308, 419-21	33.3	1971
546	Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis. *Nature*, **2011**, 476, 214-9	50.4	1948
545	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
544	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
543	Common variants at MS4A4/MS4A6E, CD2AP, CD33 and EPHA1 are associated with late-onset Alzheimer's disease. *Nature Genetics*, **2011**, 43, 436-41	36.3	1367
542	Risk alleles for multiple sclerosis identified by a genomewide study. *New England Journal of Medicine*, **2007**, 357, 851-62	59.2	1327
541	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **2007**, 39, 319-28	36.3	1083
540	Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis. *Nature Genetics*, **2013**, 45, 1353-60	36.3	934
539	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
538	Mutations in UBQLN2 cause dominant X-linked juvenile and adult-onset ALS and ALS/dementia. *Nature*, **2011**, 477, 211-5	50.4	846
537	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. *Nature Genetics*, **2016**, 48, 134-43	36.3	769
536	Common genetic variants on 5p14.1 associate with autism spectrum disorders. *Nature*, **2009**, 459, 528-33	50.4	760
535	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *American Journal of Human Genetics*, **2014**, 94, 677-94	11	635
534	Meta-analysis of genome scans and replication identify CD6, IRF8 and TNFRSF1A as new multiple sclerosis susceptibility loci. *Nature Genetics*, **2009**, 41, 776-82	36.3	621

533	Seven new loci associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 433-9, 439e1-2	36.3	577
532	Systematic comparison of phenome-wide association study of electronic medical record data and genome-wide association study data. *Nature Biotechnology*, **2013**, 31, 1102-10	44.5	555
531	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, 1322-8	10.2	512
530	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. *Nature Genetics*, **2017**, 49, 1373-1384	36.3	508
529	Interleukin 7 receptor alpha chain (IL7R) shows allelic and functional association with multiple sclerosis. *Nature Genetics*, **2007**, 39, 1083-91	36.3	506
528	The Electronic Medical Records and Genomics (eMERGE) Network: past, present, and future. *Genetics in Medicine*, **2013**, 15, 761-71	8.1	484
527	Isolation of a novel gene underlying Batten disease, CLN3. The International Batten Disease Consortium. *Cell*, **1995**, 82, 949-57	56.2	484
526	Mitochondrial polymorphisms significantly reduce the risk of Parkinson disease. *American Journal of Human Genetics*, **2003**, 72, 804-11	11	458
525	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443
524	Genome-wide association study identifies a new breast cancer susceptibility locus at 6q25.1. *Nature Genetics*, **2009**, 41, 324-8	36.3	434
523	Genetic variants near TIMP3 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, 7401-6	11.5	417
522	Genetic linkage of bilateral acoustic neurofibromatosis to a DNA marker on chromosome 22. *Nature*, **1987**, 329, 246-8	50.4	413
521	The PhenX Toolkit: get the most from your measures. *American Journal of Epidemiology*, **2011**, 174, 253-60	5.8	397
520	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
519	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, 1381-4	59.2	355
518	Meta-analysis confirms CR1, CLU, and PICALM as alzheimer disease risk loci and reveals interactions with APOE genotypes. *Archives of Neurology*, **2010**, 67, 1473-84		330
517	Multiple sclerosis genomic map implicates peripheral immune cells and microglia in susceptibility. *Science*, **2019**, 365,	33.3	309
516	Cigarette smoking strongly modifies the association of LOC387715 and age-related macular degeneration. *American Journal of Human Genetics*, **2006**, 78, 852-864	11	284

515	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
514	Variants in the ATP-binding cassette transporter (ABCA7), apolipoprotein E ?4,and the risk of late-onset Alzheimer disease in African Americans. *JAMA - Journal of the American Medical Association*, **2013**, 309, 1483-92	27.4	275
513	Mapping multiple sclerosis susceptibility to the HLA-DR locus in African Americans. *American Journal of Human Genetics*, **2004**, 74, 160-7	11	274
512	Robust replication of genotype-phenotype associations across multiple diseases in an electronic medical record. *American Journal of Human Genetics*, **2010**, 86, 560-72	11	264
511	Genome-wide meta-analysis identifies novel multiple sclerosis susceptibility loci. *Annals of Neurology*, **2011**, 70, 897-912	9.4	263
510	Age at onset in two common neurodegenerative diseases is genetically controlled. *American Journal of Human Genetics*, **2002**, 70, 985-93	11	262
509	GWAS of cerebrospinal fluid tau levels identifies risk variants for Alzheimer's disease. *Neuron*, **2013**, 78, 256-68	13.9	255
508	The genetic defect in familial Alzheimer's disease is not tightly linked to the amyloid beta-protein gene. *Nature*, **1987**, 329, 156-7	50.4	253
507	Heterogeneity at the HLA-DRB1 locus and risk for multiple sclerosis. *Human Molecular Genetics*, **2006**, 15, 2813-24	5.6	246
506	Cardiac sodium channel (SCN5A) variants associated with atrial fibrillation. *Circulation*, **2008**, 117, 1927-35	6.7	245
505	Analysis of European mitochondrial haplogroups with Alzheimer disease risk. *Neuroscience Letters*, **2004**, 365, 28-32	3.3	237
504	A high-density screen for linkage in multiple sclerosis. *American Journal of Human Genetics*, **2005**, 77, 454-67	11	235
503	A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. *Nature Neuroscience*, **2017**, 20, 1052-1061	25.5	228
502	Common variants at 9p21 and 8q22 are associated with increased susceptibility to optic nerve degeneration in glaucoma. *PLoS Genetics*, **2012**, 8, e1002654	6	227
501	Common genetic variants in the CLDN2 and PRSS1-PRSS2 loci alter risk for alcohol-related and sporadic pancreatitis. *Nature Genetics*, **2012**, 44, 1349-54	36.3	223
500	Genome-wide association analyses identify multiple loci associated with central corneal thickness and keratoconus. *Nature Genetics*, **2013**, 45, 155-63	36.3	222
499	Genome-wide association meta-analysis of neuropathologic features of Alzheimer's disease and related dementias. *PLoS Genetics*, **2014**, 10, e1004606	6	219
498	Class II HLA interactions modulate genetic risk for multiple sclerosis. *Nature Genetics*, **2015**, 47, 1107-1113	36.3	215

497	Genome-wide association study implicates a chromosome 12 risk locus for late-onset Alzheimer disease. *American Journal of Human Genetics*, **2009**, 84, 35-43	11	215
496	Temperature-sensitive mutations in the III-IV cytoplasmic loop region of the skeletal muscle sodium channel gene in paramyotonia congenita. *Cell*, **1992**, 68, 769-74	56.2	215
495	Genetic assessment of age-associated Alzheimer disease risk: Development and validation of a polygenic hazard score. *PLoS Medicine*, **2017**, 14, e1002258	11.6	209
494	Complete genomic screen in Parkinson disease: evidence for multiple genes. *JAMA - Journal of the American Medical Association*, **2001**, 286, 2239-44	27.4	208
493	Human gene for torsion dystonia located on chromosome 9q32-q34. *Neuron*, **1989**, 2, 1427-34	13.9	207
492	Genetics, statistics and human disease: analytical retooling for complexity. *Trends in Genetics*, **2004**, 20, 640-7	8.5	205
491	Quality control procedures for genome-wide association studies. *Current Protocols in Human Genetics*, **2011**, Chapter 1, Unit1.19	3.2	199
490	Meta-analysis of genome scans of age-related macular degeneration. *Human Molecular Genetics*, **2005**, 14, 2257-64	5.6	197
489	Linkage of recessive familial amyotrophic lateral sclerosis to chromosome 2q33-q35. *Nature Genetics*, **1994**, 7, 425-8	36.3	185
488	PheKB: a catalog and workflow for creating electronic phenotype algorithms for transportability. *Journal of the American Medical Informatics Association: JAMIA*, **2016**, 23, 1046-1052	8.6	184
487	A genome-wide association study of autism reveals a common novel risk locus at 5p14.1. *Annals of Human Genetics*, **2009**, 73, 263-73	2.2	179
486	Brain expression genome-wide association study (eGWAS) identifies human disease-associated variants. *PLoS Genetics*, **2012**, 8, e1002707	6	174
485	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, 3500-12	5.6	174
484	Prevalence of mutations in TIGR/Myocilin in patients with adult and juvenile primary open-angle glaucoma. *American Journal of Human Genetics*, **1998**, 63, 1549-52	11	173
483	Glutathione S-transferase omega-1 modifies age-at-onset of Alzheimer disease and Parkinson disease. *Human Molecular Genetics*, **2003**, 12, 3259-67	5.6	173
482	Parkin mutations and susceptibility alleles in late-onset Parkinson's disease. *Annals of Neurology*, **2003**, 53, 624-9	9.4	172
481	Mutations in a novel CLN6-encoded transmembrane protein cause variant neuronal ceroid lipofuscinosis in man and mouse. *American Journal of Human Genetics*, **2002**, 70, 324-35	11	172
480	Genome-wide analysis of multi-ancestry cohorts identifies new loci influencing intraocular pressure and susceptibility to glaucoma. *Nature Genetics*, **2014**, 46, 1126-1130	36.3	171

479	Incorporating language phenotypes strengthens evidence of linkage to autism. *American Journal of Medical Genetics Part A*, **2001**, 105, 539-547		169
478	Neonatal pulmonary hypertension--urea-cycle intermediates, nitric oxide production, and carbamoyl-phosphate synthetase function. *New England Journal of Medicine*, **2001**, 344, 1832-8	59.2	169
477	Clustering of autoimmune diseases in families with a high-risk for multiple sclerosis: a descriptive study. *Lancet Neurology, The*, **2006**, 5, 924-31	24.1	166
476	Construction of a GT polymorphism map of human 9q. *Genomics*, **1992**, 12, 229-40	4.3	165
475	Protective effect of complement factor B and complement component 2 variants in age-related macular degeneration. *Human Molecular Genetics*, **2007**, 16, 1986-92	5.6	161
474	The role of the CD58 locus in multiple sclerosis. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 5264-9	11.5	160
473	Genome-wide association analysis identifies TXNRD2, ATXN2 and FOXC1 as susceptibility loci for primary open-angle glaucoma. *Nature Genetics*, **2016**, 48, 189-94	36.3	159
472	Exome sequencing of extended families with autism reveals genes shared across neurodevelopmental and neuropsychiatric disorders. *Molecular Autism*, **2014**, 5, 1	6.5	158
471	Functional candidate genes in age-related macular degeneration: significant association with VEGF, VLDLR, and LRP6. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 329-35		158
470	A highly polymorphic locus very tightly linked to the Huntington's disease gene. *Nature*, **1988**, 332, 734-6	50.4	155
469	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, 174-89	11	154
468	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
467	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimerls and Dementia*, **2015**, 11, 658-71	1.2	146
466	Evidence for polygenic susceptibility to multiple sclerosis--the shape of things to come. *American Journal of Human Genetics*, **2010**, 86, 621-5	11	146
465	Linkage disequilibrium at the Angelman syndrome gene UBE3A in autism families. *Genomics*, **2001**, 77, 105-13	4.3	145
464	Sex-Specific Association of Apolipoprotein E With Cerebrospinal Fluid Levels of Tau. *JAMA Neurology*, **2018**, 75, 989-998	17.2	142
463	Common variants near ABCA1, AFAP1 and GMDS confer risk of primary open-angle glaucoma. *Nature Genetics*, **2014**, 46, 1120-1125	36.3	141
462	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, 849-59	3.8	141

461	A second major histocompatibility complex susceptibility locus for multiple sclerosis. *Annals of Neurology*, **2007**, 61, 228-36	9.4	140
460	Neurofibromatosis 2: clinical and DNA linkage studies of a large kindred. *New England Journal of Medicine*, **1988**, 319, 278-83	59.2	140
459	A noise-reduction GWAS analysis implicates altered regulation of neurite outgrowth and guidance in autism. *Molecular Autism*, **2011**, 2, 1	6.5	139
458	Meta-analysis of the association between variants in SORL1 and Alzheimer disease. *Archives of Neurology*, **2011**, 68, 99-106		135
457	Evaluation of copy number variations reveals novel candidate genes in autism spectrum disorder-associated pathways. *Human Molecular Genetics*, **2012**, 21, 3513-23	5.6	135
456	Genome- and phenome-wide analyses of cardiac conduction identifies markers of arrhythmia risk. *Circulation*, **2013**, 127, 1377-85	16.7	133
455	Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma in Caucasians from the USA. *Human Molecular Genetics*, **2011**, 20, 4707-13	5.6	133
454	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. *Nature Genetics*, **1993**, 4, 160-4	36.3	133
453	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
452	Identification of a rare coding variant in complement 3 associated with age-related macular degeneration. *Nature Genetics*, **2013**, 45, 1375-9	36.3	130
451	Whole-exome sequencing links a variant in DHDDS to retinitis pigmentosa. *American Journal of Human Genetics*, **2011**, 88, 201-6	11	130
450	Effects of multiple genetic loci on age at onset in late-onset Alzheimer disease: a genome-wide association study. *JAMA Neurology*, **2014**, 71, 1394-404	17.2	129
449	Rapidly progressive Alzheimer's disease features distinct structures of amyloid-□*Brain*, **2015**, 138, 1009-22	11.2	128
448	Novel late-onset Alzheimer disease loci variants associate with brain gene expression. *Neurology*, **2012**, 79, 221-8	6.5	124
447	Localization of the Huntington's disease gene to a small segment of chromosome 4 flanked by D4S10 and the telomere. *Cell*, **1987**, 50, 565-71	56.2	124
446	SORL1 is genetically associated with late-onset Alzheimer's disease in Japanese, Koreans and Caucasians. *PLoS ONE*, **2013**, 8, e58618	3.7	122
445	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	36.3	122
444	Cu/Zn superoxide dismutase activity in familial and sporadic amyotrophic lateral sclerosis. *Journal of Neurochemistry*, **1994**, 62, 384-7	6	119

443	A linkage disequilibrium map of the 1-Mb 15q12 GABA(A) receptor subunit cluster and association to autism. *American Journal of Medical Genetics Part A*, **2004**, 131B, 51-9		119
442	Assessment of the genetic variance of late-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 41, 200.e13-200.e20	5.6	119
441	A pooled case-control study of the apolipoprotein E (APOE) gene in age-related maculopathy. *Ophthalmic Genetics*, **2002**, 23, 209-23	1.2	116
440	Mitochondrial haplogroups and peripheral neuropathy during antiretroviral therapy: an adult AIDS clinical trials group study. *Aids*, **2005**, 19, 1341-9	3.5	114
439	Exceptionally low likelihood of Alzheimer's dementia in APOE2 homozygotes from a 5,000-person neuropathological study. *Nature Communications*, **2020**, 11, 667	17.4	113
438	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
437	Investigation of autism and GABA receptor subunit genes in multiple ethnic groups. *Neurogenetics*, **2006**, 7, 167-74	3	110
436	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	110
435	C3 R102G polymorphism increases risk of age-related macular degeneration. *Human Molecular Genetics*, **2008**, 17, 1821-4	5.6	109
434	Genetic susceptibility to Alzheimer disease. *Trends in Genetics*, **1995**, 11, 504-8	8.5	109
433	Genome-wide association study identifies four novel loci associated with Alzheimer's endophenotypes and disease modifiers. *Acta Neuropathologica*, **2017**, 133, 839-856	14.3	107
432	Identification of genomic predictors of atrioventricular conduction: using electronic medical records as a tool for genome science. *Circulation*, **2010**, 122, 2016-21	16.7	107
431	Replication analysis identifies TYK2 as a multiple sclerosis susceptibility factor. *European Journal of Human Genetics*, **2009**, 17, 1309-13	5.3	107
430	An alpha-2-macroglobulin insertion-deletion polymorphism in Alzheimer disease. *Nature Genetics*, **1999**, 22, 19-22	36.3	107
429	Transethnic genome-wide scan identifies novel Alzheimer's disease loci. *Alzheimerls and Dementia*, **2017**, 13, 727-738	1.2	106
428	Association of polymorphisms in the apolipoprotein E region with susceptibility to and progression of multiple sclerosis. *American Journal of Human Genetics*, **2002**, 70, 708-17	11	106
427	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	15.1	106
426	Recurrent tissue-specific mtDNA mutations are common in humans. *PLoS Genetics*, **2013**, 9, e1003929	6	105

425	Identification of two novel loci for dominantly inherited familial amyotrophic lateral sclerosis. *American Journal of Human Genetics*, **2003**, 73, 397-403	11	105
424	Distribution of WDR36 DNA sequence variants in patients with primary open-angle glaucoma. *Investigative Ophthalmology and Visual Science*, **2006**, 47, 2542-6		104
423	Polygenic Overlap Between C-Reactive Protein, Plasma Lipids, and Alzheimer Disease. *Circulation*, **2015**, 131, 2061-2069	16.7	100
422	A comparison of neurological, metabolic, structural, and genetic evaluations in persons at risk for Huntington's disease. *Annals of Neurology*, **1990**, 28, 614-21	9.4	100
421	A rare mutation in UNC5C predisposes to late-onset Alzheimer's disease and increases neuronal cell death. *Nature Medicine*, **2014**, 20, 1452-7	50.5	97
420	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, 1788-92		96
419	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
418	Endothelial nitric oxide synthase gene variants and primary open-angle glaucoma: interactions with sex and postmenopausal hormone use **2010**, 51, 971-9		95
417	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, 953-62	5.6	91
416	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	91
415	Imputation and quality control steps for combining multiple genome-wide datasets. *Frontiers in Genetics*, **2014**, 5, 370	4.5	90
414	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
413	Recombination events suggest potential sites for the Huntington's disease gene. *Neuron*, **1989**, 3, 183-90	13.9	88
412	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, 856-63	7.3	87
411	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
410	Missense variant in TREML2 protects against Alzheimer's disease. *Neurobiology of Aging*, **2014**, 35, 1510.e19-26	5.1	84
409	A common variant near TGFBR3 is associated with primary open angle glaucoma. *Human Molecular Genetics*, **2015**, 24, 3880-92	5.6	84
408	Two rare AKAP9 variants are associated with Alzheimer's disease in African Americans. *Alzheimerls and Dementia*, **2014**, 10, 609-618.e11	1.2	83

407	Drug transporter and metabolizing enzyme gene variants and nonnucleoside reverse-transcriptase inhibitor hepatotoxicity. *Clinical Infectious Diseases*, **2006**, 43, 779-82	11.6	83
406	Comprehensive search for Alzheimer disease susceptibility loci in the APOE region. *Archives of Neurology*, **2012**, 69, 1270-9		81
405	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, 438-453	5.6	80
404	Mitochondrial DNA polymorphism A4917G is independently associated with age-related macular degeneration. *PLoS ONE*, **2008**, 3, e2091	3.7	80
403	Analysis of the Stargardt disease gene (ABCR) in age-related macular degeneration. *Ophthalmology* , **1999**, 106, 1531-6	7.3	80
402	Genetic linkage of autosomal dominant juvenile glaucoma to 1q21-q31 in three affected pedigrees. *Genomics*, **1994**, 21, 299-303	4.3	80
401	Genome-wide association study and meta-analysis of intraocular pressure. *Human Genetics*, **2014**, 133, 41-57	6.3	79
400	SORCS1 alters amyloid precursor protein processing and variants may increase Alzheimer's disease risk. *Annals of Neurology*, **2011**, 69, 47-64	9.4	79
399	Multitrait analysis of glaucoma identifies new risk loci and enables polygenic prediction of disease susceptibility and progression. *Nature Genetics*, **2020**, 52, 160-166	36.3	78
398	Association of MAPT haplotypes with Alzheimer's disease risk and MAPT brain gene expression levels. *Alzheimerls Research and Therapy*, **2014**, 6, 39	9	78
397	Deletion of CFHR3 and CFHR1 genes in age-related macular degeneration. *Human Molecular Genetics*, **2008**, 17, 971-7	5.6	78
396	Lack of association of mutations in optineurin with disease in patients with adult-onset primary open-angle glaucoma. *JAMA Ophthalmology*, **2003**, 121, 1181-3		77
395	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	6	76
394	Association between apolipoprotein E genotype and Alzheimer disease in African American subjects. *Archives of Neurology*, **2002**, 59, 594-600		76
393	Novel mutations detected in the TSC2 gene from both sporadic and familial TSC patients. *Human Molecular Genetics*, **1996**, 5, 249-56	5.6	76
392	Early adult-onset POAG linked to 15q11-13 using ordered subset analysis. *Investigative Ophthalmology and Visual Science*, **2005**, 46, 2002-5		74
391	A genetic linkage map of the long arm of human chromosome 22. *Genomics*, **1989**, 4, 1-6	4.3	74
390	Association of CAV1/CAV2 genomic variants with primary open-angle glaucoma overall and by gender and pattern of visual field loss. *Ophthalmology*, **2014**, 121, 508-16	7.3	73

389	Localization of age-related macular degeneration-associated ARMS2 in cytosol, not mitochondria **2009**, 50, 3084-90		73
388	Distribution of optineurin sequence variations in an ethnically diverse population of low-tension glaucoma patients from the United States. *Journal of Glaucoma*, **2006**, 15, 358-63	2.1	73
387	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. *Nature Genetics*, **2017**, 49, 993-1004	36.3	72
386	Genome-wide association study identifies seven novel susceptibility loci for primary open-angle glaucoma. *Human Molecular Genetics*, **2018**, 27, 1486-1496	5.6	72
385	Assessing the accuracy of observer-reported ancestry in a biorepository linked to electronic medical records. *Genetics in Medicine*, **2010**, 12, 648-50	8.1	72
384	Independent effects of complement factor H Y402H polymorphism and cigarette smoking on risk of age-related macular degeneration. *Ophthalmology*, **2007**, 114, 1151-6	7.3	72
383	Low-Frequency and Rare-Coding Variation Contributes to Multiple Sclerosis Risk. *Cell*, **2018**, 175, 1679-1687.e7	36.2	72
382	Meta-analysis of genome-wide association studies identifies novel loci that influence cupping and the glaucomatous process. *Nature Communications*, **2014**, 5, 4883	17.4	71
381	GATA2 is associated with familial early-onset coronary artery disease. *PLoS Genetics*, **2006**, 2, e139	6	71
380	The CEPH consortium primary linkage map of human chromosome 10. *Genomics*, **1990**, 6, 393-412	4.3	70
379	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, 170-9	4.1	69
378	Familial Alzheimer's disease: progress and problems. *Neurobiology of Aging*, **1989**, 10, 417-25	5.6	67
377	eMERGEing progress in genomics-the first seven years. *Frontiers in Genetics*, **2014**, 5, 184	4.5	65
376	The expanding role of MBD genes in autism: identification of a MECP2 duplication and novel alterations in MBD5, MBD6, and SETDB1. *Autism Research*, **2012**, 5, 385-97	5.1	65
375	Apolipoprotein E genotype in patients with Alzheimer's disease: implications for the risk of dementia among relatives. *Annals of Neurology*, **1995**, 38, 797-808	9.4	65
374	Characterization of a translocation within the von Recklinghausen neurofibromatosis region of chromosome 17. *Genomics*, **1989**, 5, 245-9	4.3	63
373	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	4.9	61
372	Copy number variants in extended autism spectrum disorder families reveal candidates potentially involved in autism risk. *PLoS ONE*, **2011**, 6, e26049	3.7	61

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370	Migraine with aura susceptibility locus on chromosome 19p13 is distinct from the familial hemiplegic migraine locus. *Genomics*, **2001**, 78, 150-4	4.3	61
369	C9ORF72 intermediate repeat copies are a significant risk factor for Parkinson disease. *Annals of Human Genetics*, **2013**, 77, 351-63	2.2	60
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