Benjamin M Neale

List of Publications by Year
in descending order

245 papers	118,487 citations	1701 104 h-index	872 243 g-index
388 all docs	388 docs citations	388 times ranked	106684 citing authors

#	Article	IF	Citations
1	PLINK: A Tool Set for Whole-Genome Association and Population-Based Linkage Analyses. American Journal of Human Genetics, 2007, 81, 559-575.	2.6	26,761
2	Analysis of protein-coding genetic variation in 60,706 humans. Nature, 2016, 536, 285-291.	13.7	9,051
3	The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2020, 581, 434-443.	13.7	6,140
4	LD Score regression distinguishes confounding from polygenicity in genome-wide association studies. Nature Genetics, 2015, 47, 291-295.	9.4	3,905
5	Detection of widespread horizontal pleiotropy in causal relationships inferred from Mendelian randomization between complex traits and diseases. Nature Genetics, 2018, 50, 693-698.	9.4	3,593
6	An atlas of genetic correlations across human diseases and traits. Nature Genetics, 2015, 47, 1236-1241.	9.4	3,145
7	Clonal Hematopoiesis and Blood-Cancer Risk Inferred from Blood DNA Sequence. New England Journal of Medicine, 2014, 371, 2477-2487.	13.9	2,669
8	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	13.7	2,254
9	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. Nature Genetics, 2013, 45, 984-994.	9.4	2,067
10	Partitioning heritability by functional annotation using genome-wide association summary statistics. Nature Genetics, 2015, 47, 1228-1235.	9.4	2,045
11	Clinical use of current polygenic risk scores may exacerbate health disparities. Nature Genetics, 2019, 51, 584-591.	9.4	1,664
12	Patterns and rates of exonic de novo mutations in autism spectrum disorders. Nature, 2012, 485, 242-245.	13.7	1,597
13	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. Nature Genetics, 2019, 51, 63-75.	9.4	1,594
14	Identification of common genetic risk variants for autism spectrum disorder. Nature Genetics, 2019, 51, 431-444.	9.4	1,538
15	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	13.5	1,422
16	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. Nature Genetics, 2013, 45, 1150-1159.	9.4	1,395
17	Efficient Bayesian mixed-model analysis increases association power in large cohorts. Nature Genetics, 2015, 47, 284-290.	9.4	1,285
18	Genome-wide association study identifies 30 loci associated with bipolar disorder. Nature Genetics, 2019, 51, 793-803.	9.4	1,191

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19	Human Demographic History Impacts Genetic Risk Prediction across Diverse Populations. American Journal of Human Genetics, 2017, 100, 635-649.	2.6	1,120
20	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. American Journal of Human Genetics, 2015, 97, 576-592.	2.6	1,098
21	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
22	A mega-analysis of genome-wide association studies for major depressive disorder. Molecular Psychiatry, 2013, 18, 497-511.	4.1	1,002
23	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	13.7	952
24	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
25	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	13.7	929
26	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. Science, 2018, 359, 693-697.	6.0	851
27	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. Nature Genetics, 2017, 49, 27-35.	9.4	838
28	LD Hub: a centralized database and web interface to perform LD score regression that maximizes the potential of summary level GWAS data for SNP heritability and genetic correlation analysis. Bioinformatics, 2017, 33, 272-279.	1.8	822
29	Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types. Nature Genetics, 2018, 50, 621-629.	9.4	807
30	A global overview of pleiotropy and genetic architecture in complex traits. Nature Genetics, 2019, 51, 1339-1348.	9.4	774
31	Common variants associated with plasma triglycerides and risk for coronary artery disease. Nature Genetics, 2013, 45, 1345-1352.	9.4	754
32	Multi-trait analysis of genome-wide association summary statistics using MTAG. Nature Genetics, 2018, 50, 229-237.	9.4	700
33	Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. Nature Genetics, 2011, 43, 1066-1073.	9.4	698
34	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	13.7	640
35	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. Nature Genetics, 2021, 53, 817-829.	9.4	629
36	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614

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37	The Future of Association Studies: Gene-Based Analysis and Replication. American Journal of Human Genetics, 2004, 75, 353-362.	2.6	598
38	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. Nature Genetics, 2013, 45, 501-512.	9.4	578
39	Genome-wide association identifies multiple ulcerative colitis susceptibility loci. Nature Genetics, 2010, 42, 332-337.	9.4	572
40	Searching for missing heritability: Designing rare variant association studies. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E455-64.	3.3	570
41	Partitioning Heritability of Regulatory and Cell-Type-Specific Variants across 11 Common Diseases. American Journal of Human Genetics, 2014, 95, 535-552.	2.6	569
42	Pervasive Sharing of Genetic Effects in Autoimmune Disease. PLoS Genetics, 2011, 7, e1002254.	1.5	540
43	Sequencing Chromosomal Abnormalities Reveals Neurodevelopmental Loci that Confer Risk across Diagnostic Boundaries. Cell, 2012, 149, 525-537.	13.5	534
44	Testing for an Unusual Distribution of Rare Variants. PLoS Genetics, 2011, 7, e1001322.	1.5	530
45	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. Nature Genetics, 2016, 48, 856-866.	9.4	520
46	Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. Human Molecular Genetics, 2009, 18, 472-481.	1.4	512
47	Two independent alleles at 6q23 associated with risk of rheumatoid arthritis. Nature Genetics, 2007, 39, 1477-1482.	9.4	497
48	Genetics of blood lipids among ~300,000 multi-ethnic participants of the Million Veteran Program. Nature Genetics, 2018, 50, 1514-1523.	9.4	497
49	Transancestral GWAS of alcohol dependence reveals common genetic underpinnings with psychiatric disorders. Nature Neuroscience, 2018, 21, 1656-1669.	7.1	490
50	Common variants at CD40 and other loci confer risk of rheumatoid arthritis. Nature Genetics, 2008, 40, 1216-1223.	9.4	476
51	Practical aspects of imputation-driven meta-analysis of genome-wide association studies. Human Molecular Genetics, 2008, 17, R122-R128.	1.4	475
52	Comparative genetic architectures of schizophrenia in East Asian and European populations. Nature Genetics, 2019, 51, 1670-1678.	9.4	440
53	Contrasting genetic architectures of schizophrenia and other complex diseases using fast variance-components analysis. Nature Genetics, 2015, 47, 1385-1392.	9.4	431
54	Meta-Analysis of Genome-Wide Association Studies of Attention-Deficit/Hyperactivity Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 884-897.	0.3	423

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55	Genome-wide association study of advanced age-related macular degeneration identifies a role of the hepatic lipase gene (LIPC). Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7395-7400.	3.3	406
56	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. Nature Genetics, 2018, 50, 538-548.	9.4	406
57	Variation in complement factor 3 is associated with risk of age-related macular degeneration. Nature Genetics, 2007, 39, 1200-1201.	9.4	405
58	Linkage disequilibriumâ€“dependent architecture of human complex traits shows action of negative selection. Nature Genetics, 2017, 49, 1421-1427.	9.4	400
59	Genome-wide association studies in ADHD. Human Genetics, 2009, 126, 13-50.	1.8	374
60	International meta-analysis of PTSD genome-wide association studies identifies sex- and ancestry-specific genetic risk loci. Nature Communications, 2019, 10, 4558.	5.8	363
61	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. Nature Genetics, 2015, 47, 1294-1303.	9.4	357
62	Large-scale genome-wide meta-analysis of polycystic ovary syndrome suggests shared genetic architecture for different diagnosis criteria. PLoS Genetics, 2018, 14, e1007813.	1.5	341
63	Exome sequencing and the genetic basis of complex traits. Nature Genetics, 2012, 44, 623-630.	9.4	340
64	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	9.4	338
65	Genomeâ€wide association scan of quantitative traits for attention deficit hyperactivity disorder identifies novel associations and confirms candidate gene associations. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1345-1354.	1.1	335
66	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. Nature Genetics, 2016, 48, 552-555.	9.4	326
67	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	13.7	326
68	Variation near complement factor I is associated with risk of advanced AMD. European Journal of Human Genetics, 2009, 17, 100-104.	1.4	324
69	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. Nature Genetics, 2017, 49, 504-510.	9.4	298
70	Polygenic and clinical risk scores and their impact on age at onset and prediction of cardiometabolic diseases and common cancers. Nature Medicine, 2020, 26, 549-557.	15.2	281
71	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. ELife, 2019, 8, .	2.8	276
72	Predicting Polygenic Risk of Psychiatric Disorders. Biological Psychiatry, 2019, 86, 97-109.	0.7	252

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73	Exome array analysis identifies new loci and low-frequency variants influencing insulin processing and secretion. Nature Genetics, 2013, 45, 197-201.	9.4	247
74	Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior. Science, 2019, 365, .	6.0	245
75	Rare Complete Knockouts in Humans: Population Distribution and Significant Role in Autism Spectrum Disorders. Neuron, 2013, 77, 235-242.	3.8	242
76	Interrogating the Genetic Determinants of Touretteâ€™s Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. American Journal of Psychiatry, 2019, 176, 217-227.	4.0	242
77	Partitioning the Heritability of Tourette Syndrome and Obsessive Compulsive Disorder Reveals Differences in Genetic Architecture. PLoS Genetics, 2013, 9, e1003864.	1.5	241
78	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. Nature Genetics, 2012, 44, 631-635.	9.4	239
79	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	2.6	237
80	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. Nature Genetics, 2018, 50, 727-736.	9.4	235
81	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. Science, 2018, 362, .	6.0	234
82	The statistical properties of gene-set analysis. Nature Reviews Genetics, 2016, 17, 353-364.	7.7	230
83	Genomeâ€wide association scan of attention deficit hyperactivity disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1337-1344.	1.1	228
84	Comparison of methods that use whole genome data to estimate the heritability and genetic architecture of complex traits. Nature Genetics, 2018, 50, 737-745.	9.4	205
85	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. Nature Neuroscience, 2016, 19, 420-431.	7.1	204
86	RICOPILI: Rapid Imputation for COnsortias PIpeLIne. Bioinformatics, 2020, 36, 930-933.	1.8	201
87	zCall: a rare variant caller for array-based genotyping. Bioinformatics, 2012, 28, 2543-2545.	1.8	195
88	Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89.	13.7	194
89	Whole exome sequencing study identifies novel rare and common Alzheimerâ€™s-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	4.1	191
90	Phenome-wide heritability analysis of the UK Biobank. PLoS Genetics, 2017, 13, e1006711.	1.5	191

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91	A genome-wide association study of shared risk across psychiatric disorders implicates gene regulation during fetal neurodevelopment. Nature Neuroscience, 2019, 22, 353-361.	7.1	173
92	Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects. Nature Communications, 2018, 9, 4038.	5.8	166
93	Genome-wide Association Studies of Posttraumatic Stress Disorder in 2 Cohorts of US Army Soldiers. JAMA Psychiatry, 2016, 73, 695.	6.0	158
94	Polygenic transmission and complex neuro developmental network for attention deficit hyperactivity disorder: Genomeâ€wide association study of both common and rare variants. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 419-430.	1.1	157
95	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. Neuron, 2017, 94, 486-499.e9.	3.8	155
96	A synthetic-diploid benchmark for accurate variant-calling evaluation. Nature Methods, 2018, 15, 595-597.	9.0	154
97	Clozapine-induced agranulocytosis is associated with rare HLA-DQB1 and HLA-B alleles. Nature Communications, 2014, 5, 4757.	5.8	153
98	Case-Control Genome-Wide Association Study of Attention-Deficit/Hyperactivity Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 906-920.	0.3	150
99	Autism spectrum disorder and attention deficit hyperactivity disorder have a similar burden of rare protein-truncating variants. Nature Neuroscience, 2019, 22, 1961-1965.	7.1	148
100	A Genetic Investigation of Sex Bias in the Prevalence of Attention-Deficit/Hyperactivity Disorder. Biological Psychiatry, 2018, 83, 1044-1053.	0.7	146
101	Dynamic incorporation of multiple in silico functional annotations empowers rare variant association analysis of large whole-genome sequencing studies at scale. Nature Genetics, 2020, 52, 969-983.	9.4	146
102	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. Nature Communications, 2018, 9, 3391.	5.8	140
103	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. Neuron, 2017, 94, 1101-1111.e7.	3.8	137
104	Analysis of Rare, Exonic Variation amongst Subjects with Autism Spectrum Disorders and Population Controls. PLoS Genetics, 2013, 9, e1003443.	1.5	133
105	High Loading of Polygenic Risk for ADHD in Children With Comorbid Aggression. American Journal of Psychiatry, 2013, 170, 909-916.	4.0	127
106	Autism spectrum disorder severity reflects the average contribution of de novo and familial influences. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15161-15165.	3.3	125
107	Exome sequencing in schizophrenia-affected parentâ€“offspring trios reveals risk conferred by protein-coding de novo mutations. Nature Neuroscience, 2020, 23, 185-193.	7.1	125
108	Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. Nature Genetics, 2021, 53, 195-204.	9.4	125

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109	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts. Nature Genetics, 2020, 52, 634-639.	9.4	124
110	Genetic analyses identify widespread sex-differential participation bias. Nature Genetics, 2021, 53, 663-671.	9.4	124
111	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	7.1	122
112	Estimation of Genetic Correlation via Linkage Disequilibrium Score Regression and Genomic Restricted Maximum Likelihood. American Journal of Human Genetics, 2018, 102, 1185-1194.	2.6	119
113	Cross-Disorder Genome-Wide Analyses Suggest a Complex Genetic Relationship Between Touretteâ€™s Syndrome and OCD. American Journal of Psychiatry, 2015, 172, 82-93.	4.0	117
114	Whole-genome analyses of whole-brain data: working within an expanded search space. Nature Neuroscience, 2014, 17, 791-800.	7.1	112
115	A Genome-Wide Association Meta-Analysis of Attention-Deficit/Hyperactivity Disorder Symptoms in Population-Based Pediatric Cohorts. Journal of the American Academy of Child and Adolescent Psychiatry, 2016, 55, 896-905.e6.	0.3	112
116	Copy Number Variation in Obsessive-Compulsive Disorder and Tourette Syndrome: A Cross-Disorder Study. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 910-919.	0.3	111
117	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. Nature Medicine, 2021, 27, 1012-1024.	15.2	109
118	Conduct disorder and ADHD: Evaluation of conduct problems as a categorical and quantitative trait in the international multicentre ADHD genetics study. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1369-1378.	1.1	106
119	Common body mass index-associated variants confer risk of extreme obesity. Human Molecular Genetics, 2009, 18, 3502-3507.	1.4	106
120	Problems with Using Polygenic Scores to Select Embryos. New England Journal of Medicine, 2021, 385, 78-86.	13.9	105
121	Spatiotemporal profile of postsynaptic interactomes integrates components of complex brain disorders. Nature Neuroscience, 2017, 20, 1150-1161.	7.1	104
122	Joint Contributions of Rare Copy Number Variants and Common SNPs to Risk for Schizophrenia. American Journal of Psychiatry, 2019, 176, 29-35.	4.0	104
123	A Comparison of Ten Polygenic Score Methods for Psychiatric Disorders Applied Across Multiple Cohorts. Biological Psychiatry, 2021, 90, 611-620.	0.7	103
124	Genetic Differences in the Immediate Transcriptome Response to Stress Predict Risk-Related Brain Function and Psychiatric Disorders. Neuron, 2015, 86, 1189-1202.	3.8	102
125	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	2.6	102
126	Prevalence of rearrangements in the 22q11.2 region and population-based risk of neuropsychiatric and developmental disorders in a Danish population: a case-cohort study. Lancet Psychiatry,the, 2018, 5, 573-580.	3.7	102

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127	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. Nature Neuroscience, 2019, 22, 1966-1974.	7.1	101
128	Inherited myeloproliferative neoplasm risk affects haematopoietic stem cells. Nature, 2020, 586, 769-775.	13.7	101
129	Getting genetic ancestry right for science and society. Science, 2022, 376, 250-252.	6.0	93
130	De Novo Sequence and Copy Number Variants Are Strongly Associated with Tourette Disorder and Implicate Cell Polarity in Pathogenesis. Cell Reports, 2018, 24, 3441-3454.e12.	2.9	91
131	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. Nature Neuroscience, 2016, 19, 1563-1565.	7.1	90
132	Genome-Wide Association Studies in an Isolated Founder Population from the Pacific Island of Kosrae. PLoS Genetics, 2009, 5, e1000365.	1.5	89
133	Large-Scale trans -eQTLs Affect Hundreds of Transcripts and Mediate Patterns of Transcriptional Co-regulation. American Journal of Human Genetics, 2017, 100, 581-591.	2.6	86
134	Body dissatisfaction and drive for thinness in young adult twins. International Journal of Eating Disorders, 2005, 37, 188-199.	2.1	84
135	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. Nature Communications, 2018, 9, 2606.	5.8	79
136	Genome-wide association study identifies 48 common genetic variants associated with handedness. Nature Human Behaviour, 2021, 5, 59-70.	6.2	79
137	Functional Architectures of Local and Distal Regulation of Gene Expression in Multiple Human Tissues. American Journal of Human Genetics, 2017, 100, 605-616.	2.6	76
138	The Structure of Perfectionism: A Twin Study. Behavior Genetics, 2004, 34, 483-494.	1.4	75
139	Genetic Markers of ADHD-Related Variations in Intracranial Volume. American Journal of Psychiatry, 2019, 176, 228-238.	4.0	68
140	Evaluating the cardiovascular safety of sclerostin inhibition using evidence from meta-analysis of clinical trials and human genetics. Science Translational Medicine, 2020, 12, .	5.8	68
141	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. PLoS Genetics, 2018, 14, e1007329.	1.5	66
142	Phenotypic extremes in rare variant study designs. European Journal of Human Genetics, 2016, 24, 924-930.	1.4	65
143	Exome sequencing in bipolar disorder identifies AKAP11 as a risk gene shared with schizophrenia. Nature Genetics, 2022, 54, 541-547.	9.4	65
144	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. Neuron, 2018, 98, 743-753.e4.	3.8	63

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145	Genetic analysis of schizophrenia and bipolar disorder reveals polygenicity but also suggests new directions for molecular interrogation. Current Opinion in Neurobiology, 2015, 30, 131-138.	2.0	61
146	Sex-Dependent Shared and Nonshared Genetic Architecture Across Mood and Psychotic Disorders. Biological Psychiatry, 2022, 91, 102-117.	0.7	61
147	Attention-deficit/hyperactivity disorder and lifetime cannabis use: genetic overlap and causality. Molecular Psychiatry, 2020, 25, 2493-2503.	4.1	59
148	Haplotype Sharing Provides Insights into Fine-Scale Population History and Disease in Finland. American Journal of Human Genetics, 2018, 102, 760-775.	2.6	57
149	Associations of CFHR1â€“CFHR3 deletion and a CFH SNP to age-related macular degeneration are not independent. Nature Genetics, 2010, 42, 553-555.	9.4	55
150	An Excess of Risk-Increasing Low-Frequency Variants Can Be a Signal of Polygenic Inheritance in Complex Diseases. American Journal of Human Genetics, 2014, 94, 437-452.	2.6	55
151	GWAS Identifies Risk Locus for Erectile Dysfunction and Implicates Hypothalamic Neurobiology and Diabetes in Etiology. American Journal of Human Genetics, 2019, 104, 157-163.	2.6	55
152	Genetic research in autism spectrum disorders. Current Opinion in Pediatrics, 2015, 27, 685-691.	1.0	54
153	New data and an old puzzle: the negative association between schizophrenia and rheumatoid arthritis. International Journal of Epidemiology, 2015, 44, 1706-1721.	0.9	53
154	The genetic architecture of sporadic and multiple consecutive miscarriage. Nature Communications, 2020, 11, 5980.	5.8	52
155	A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. Nature Communications, 2016, 7, 12342.	5.8	50
156	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. Nature Communications, 2020, 11, 3368.	5.8	49
157	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. American Journal of Human Genetics, 2021, 108, 656-668.	2.6	49
158	Investigating Shared Genetic Basis Across Tourette Syndrome and Comorbid Neurodevelopmental Disorders Along the Impulsivity-Compulsivity Spectrum. Biological Psychiatry, 2021, 90, 317-327.	0.7	49
159	Quantifying the Genetic Correlation between Multiple Cancer Types. Cancer Epidemiology Biomarkers and Prevention, 2017, 26, 1427-1435.	1.1	48
160	Examining Sex-Differentiated Genetic Effects Across Neuropsychiatric and Behavioral Traits. Biological Psychiatry, 2021, 89, 1127-1137.	0.7	48
161	No Evidence for Association of Autism with Rare Heterozygous Point Mutations in Contactin-Associated Protein-Like 2 (CNTNAP2), or in Other Contactin-Associated Proteins or Contactins. PLoS Genetics, 2015, 11, e1004852.	1.5	47
162	A Low-Frequency Inactivating AKT2 Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. Diabetes, 2017, 66, 2019-2032.	0.3	47

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163	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
164	Challenges and Opportunities for Developing More Generalizable Polygenic Risk Scores. Annual Review of Biomedical Data Science, 2022, 5, 293-320.	2.8	47
165	PhenoSpD: an integrated toolkit for phenotypic correlation estimation and multiple testing correction using GWAS summary statistics. GigaScience, 2018, 7, .	3.3	46
166	Exome Sequencing in Suspected Monogenic Dyslipidemias. Circulation: Cardiovascular Genetics, 2015, 8, 343-350.	5.1	45
167	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. Nature, 2021, 597, E3-E4.	13.7	45
168	Intentional Weight Loss in Young Adults: Sexâ€Specific Genetic and Environmental Effects. Obesity, 2005, 13, 745-753.	4.0	42
169	The positives, protocols, and perils of genomeâ€wide association. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1288-1294.	1.1	41
170	Linkage to Chromosome 1p36 for Attention-Deficit/Hyperactivity Disorder Traits in School and Home Settings. Biological Psychiatry, 2008, 64, 571-576.	0.7	41
171	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. Genome Research, 2018, 28, 968-974.	2.4	41
172	Discovery of rare variants for complex phenotypes. Human Genetics, 2016, 135, 625-634.	1.8	40
173	A Polygenic and Phenotypic Risk Prediction for Polycystic Ovary Syndrome Evaluated by Phenome-Wide Association Studies. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1918-1936.	1.8	40
174	Mutations in ATP13A2 (PARK9) are associated with an amyotrophic lateral sclerosis-like phenotype, implicating this locus in further phenotypic expansion. Human Genomics, 2019, 13, 19.	1.4	38
175	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	2.6	35
176	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. International Journal of Epidemiology, 2020, 49, 1022-1031.	0.9	34
177	Genome-wide risk prediction of common diseases across ancestries in one million people. Cell Genomics, 2022, 2, 100118.	3.0	34
178	Nonpaternity in Linkage Studies of Extremely Discordant Sib Pairs. American Journal of Human Genetics, 2002, 70, 526-529.	2.6	32
179	OUP accepted manuscript. Human Molecular Genetics, 2021, 30, 1521-1534.	1.4	32
180	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	2.4	31

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181	Synaptic processes and immune-related pathways implicated in Tourette syndrome. Translational Psychiatry, 2021, 11, 56.	2.4	31
182	A Note on the Parameterization of Purcellâ€™s GÂÃ—ÂE Model for Ordinal and Binary Data. Behavior Genetics, 2009, 39, 220-229.	1.4	30
183	On Genome-wide Association Studies for Family-Based Designs: An Integrative Analysis Approach Combining Ascertained Family Samples with Unselected Controls. American Journal of Human Genetics, 2010, 86, 573-580.	2.6	30
184	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. American Journal of Human Genetics, 2020, 107, 46-59.	2.6	30
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