Benjamin M Neale

List of Publications by Citations

293 papers	74,504 citations	95 h-index	272 g-index
388 ext. papers	100,963 ext. citations	18.1 avg, IF	7.3 L-index

#	Paper	IF	Citations
293	PLINK: a tool set for whole-genome association and population-based linkage analyses. *American Journal of Human Genetics*, **2007**, 81, 559-75	11	19239
292	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
291	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278
290	LD Score regression distinguishes confounding from polygenicity in genome-wide association studies. *Nature Genetics*, **2015**, 47, 291-5	36.3	2096
289	Clonal hematopoiesis and blood-cancer risk inferred from blood DNA sequence. *New England Journal of Medicine*, **2014**, 371, 2477-87	59.2	1855
288	An atlas of genetic correlations across human diseases and traits. *Nature Genetics*, **2015**, 47, 1236-41	36.3	1841
287	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
286	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
285	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
284	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. *Nature Genetics*, **2013**, 45, 1150-9	36.3	1153
283	Partitioning heritability by functional annotation using genome-wide association summary statistics. *Nature Genetics*, **2015**, 47, 1228-35	36.3	1143
282	Detection of widespread horizontal pleiotropy in causal relationships inferred from Mendelian randomization between complex traits and diseases. *Nature Genetics*, **2018**, 50, 693-698	36.3	970
281	A mega-analysis of genome-wide association studies for major depressive disorder. *Molecular Psychiatry*, **2013**, 18, 497-511	15.1	853
280	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. *Nature Genetics*, **2019**, 51, 63-75	36.3	826
279	Efficient Bayesian mixed-model analysis increases association power in large cohorts. *Nature Genetics*, **2015**, 47, 284-90	36.3	758
278	Identification of common genetic risk variants for autism spectrum disorder. *Nature Genetics*, **2019**, 51, 431-444	36.3	746
277	Clinical use of current polygenic risk scores may exacerbate health disparities. *Nature Genetics*, **2019**, 51, 584-591	36.3	711

276	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
275	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
274	Human Demographic History Impacts Genetic Risk Prediction across Diverse Populations. *American Journal of Human Genetics*, **2017**, 100, 635-649	11	665
273	Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nature Genetics*, **2019**, 51, 793-803	36.3	662
272	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656
271	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. *American Journal of Human Genetics*, **2015**, 97, 576-92	11	649
270	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
269	Deep resequencing of GWAS loci identifies independent rare variants associated with inflammatory bowel disease. *Nature Genetics*, **2011**, 43, 1066-73	36.3	584
268	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*, **2020**, 180, 568-584.e23	56.2	578
267	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. *Science*, **2018**, 359, 693-697	33.3	547
266	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	7.2	541
265	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. *Nature Genetics*, **2017**, 49, 27-35	36.3	530
264	The future of association studies: gene-based analysis and replication. *American Journal of Human Genetics*, **2004**, 75, 353-62	11	510
263	Genome-wide association identifies multiple ulcerative colitis susceptibility loci. *Nature Genetics*, **2010**, 42, 332-7	36.3	491
262	Testing for an unusual distribution of rare variants. *PLoS Genetics*, **2011**, 7, e1001322	6	465
261	Two independent alleles at 6q23 associated with risk of rheumatoid arthritis. *Nature Genetics*, **2007**, 39, 1477-82	36.3	449
260	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*, **2012**, 149, 525-37	56.2	441
259	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	11.5	437

258	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. *Nature Genetics*, **2013**, 45, 501-12	36.3	437
257	Practical aspects of imputation-driven meta-analysis of genome-wide association studies. *Human Molecular Genetics*, **2008**, 17, R122-8	5.6	423
256	Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. *Human Molecular Genetics*, **2009**, 18, 472-81	5.6	421
255	Common variants at CD40 and other loci confer risk of rheumatoid arthritis. *Nature Genetics*, **2008**, 40, 1216-23	36.3	416
254	Pervasive sharing of genetic effects in autoimmune disease. *PLoS Genetics*, **2011**, 7, e1002254	6	413
253	Partitioning heritability of regulatory and cell-type-specific variants across 11 common diseases. *American Journal of Human Genetics*, **2014**, 95, 535-52	11	411
252	Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types. *Nature Genetics*, **2018**, 50, 621-629	36.3	400
251	The mutational constraint spectrum quantified from variation in 141,456 humans		381
250	Variation in complement factor 3 is associated with risk of age-related macular degeneration. *Nature Genetics*, **2007**, 39, 1200-1	36.3	364
249	Meta-analysis of genome-wide association studies of attention-deficit/hyperactivity disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 884-97	7.2	357
248	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. *Nature Genetics*, **2016**, 48, 856-66	36.3	355
247	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, 7395-400	11.5	345
246	Multi-trait analysis of genome-wide association summary statistics using MTAG. *Nature Genetics*, **2018**, 50, 229-237	36.3	339
245	Genome-wide association studies in ADHD. *Human Genetics*, **2009**, 126, 13-50	6.3	316
244	A global overview of pleiotropy and genetic architecture in complex traits. *Nature Genetics*, **2019**, 51, 1339-1348	36.3	311
243	Exome sequencing and the genetic basis of complex traits. *Nature Genetics*, **2012**, 44, 623-30	36.3	303
242	Contrasting genetic architectures of schizophrenia and other complex diseases using fast variance-components analysis. *Nature Genetics*, **2015**, 47, 1385-92	36.3	299
241	Genome-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, 1345-54	3.5	299

240	Variation near complement factor I is associated with risk of advanced AMD. *European Journal of Human Genetics*, **2009**, 17, 100-4	5.3	290
239	Genome-wide meta-analysis identifies new susceptibility loci for migraine. *Nature Genetics*, **2013**, 45, 912-917	36.3	276
238	Genetics of blood lipids among ~300,000 multi-ethnic participants of the Million Veteran Program. *Nature Genetics*, **2018**, 50, 1514-1523	36.3	260
237	Transancestral GWAS of alcohol dependence reveals common genetic underpinnings with psychiatric disorders. *Nature Neuroscience*, **2018**, 21, 1656-1669	25.5	257
236	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. *Nature Genetics*, **2016**, 48, 552-5	36.3	238
235	Large-scale genomic analyses link reproductive aging to hypothalamic signaling, breast cancer susceptibility and BRCA1-mediated DNA repair. *Nature Genetics*, **2015**, 47, 1294-1303	36.3	226
234	A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451	50.4	223
233	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. *Nature Genetics*, **2018**, 50, 538-548	36.3	222
232	Exome array analysis identifies new loci and low-frequency variants influencing insulin processing and secretion. *Nature Genetics*, **2013**, 45, 197-201	36.3	212
231	Linkage disequilibrium-dependent architecture of human complex traits shows action of negative selection. *Nature Genetics*, **2017**, 49, 1421-1427	36.3	204
230	Refining the role of de novo protein-truncating variants in neurodevelopmental disorders by using population reference samples. *Nature Genetics*, **2017**, 49, 504-510	36.3	203
229	Genome-wide association scan of attention deficit hyperactivity disorder. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1337-44	3.5	201
228	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
227	Partitioning the heritability of Tourette syndrome and obsessive compulsive disorder reveals differences in genetic architecture. *PLoS Genetics*, **2013**, 9, e1003864	6	189
226	Comparative genetic architectures of schizophrenia in East Asian and European populations. *Nature Genetics*, **2019**, 51, 1670-1678	36.3	185
225	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. *Nature Genetics*, **2012**, 44, 631-5	36.3	184
224	Predicting Polygenic Risk of Psychiatric Disorders. *Biological Psychiatry*, **2019**, 86, 97-109	7.9	170
223	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. *ELife*, **2019**, 8,	8.9	166

222	Large-scale genome-wide meta-analysis of polycystic ovary syndrome suggests shared genetic architecture for different diagnosis criteria. *PLoS Genetics*, **2018**, 14, e1007813	6	166
221	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. *Nature Neuroscience*, **2016**, 19, 420-431	25.5	163
220	The statistical properties of gene-set analysis. *Nature Reviews Genetics*, **2016**, 17, 353-64	30.1	162
219	Mapping the human genetic architecture of COVID-19. *Nature*, **2021**,	50.4	162
218	An analytical framework for whole-genome sequence association studies and its implications for autism spectrum disorder. *Nature Genetics*, **2018**, 50, 727-736	36.3	156
217	zCall: a rare variant caller for array-based genotyping: genetics and population analysis. *Bioinformatics*, **2012**, 28, 2543-5	7.2	154
216	International meta-analysis of PTSD genome-wide association studies identifies sex- and ancestry-specific genetic risk loci. *Nature Communications*, **2019**, 10, 4558	17.4	151
215	Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior. *Science*, **2019**, 365,	33.3	139
214	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science* , **2018**, 362,	33.3	134
213	Comparison of methods that use whole genome data to estimate the heritability and genetic architecture of complex traits. *Nature Genetics*, **2018**, 50, 737-745	36.3	131
212	Case-control genome-wide association study of attention-deficit/hyperactivity disorder. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2010**, 49, 906-20	7.2	131
211	Phenome-wide heritability analysis of the UK Biobank. *PLoS Genetics*, **2017**, 13, e1006711	6	131
210	Polygenic transmission and complex neuro developmental network for attention deficit hyperactivity disorder: genome-wide association study of both common and rare variants. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2013**, 162B, 419-430	3.5	125
209	Clozapine-induced agranulocytosis is associated with rare HLA-DQB1 and HLA-B alleles. *Nature Communications*, **2014**, 5, 4757	17.4	118
208	Genome-wide Association Studies of Posttraumatic Stress Disorder in 2 Cohorts of US Army Soldiers. *JAMA Psychiatry*, **2016**, 73, 695-704	14.5	114
207	High loading of polygenic risk for ADHD in children with comorbid aggression. *American Journal of Psychiatry*, **2013**, 170, 909-16	11.9	110
206	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	50.5	109
205	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108

204	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
203	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	11	104
202	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. *Neuron*, **2017**, 94, 1101-1111.e7	13.9	103
201	Regional missense constraint improves variant deleteriousness prediction		102
200	Cross-disorder genome-wide analyses suggest a complex genetic relationship between Tourette's syndrome and OCD. *American Journal of Psychiatry*, **2015**, 172, 82-93	11.9	101
199	Whole-genome analyses of whole-brain data: working within an expanded search space. *Nature Neuroscience*, **2014**, 17, 791-800	25.5	98
198	Interrogating the Genetic Determinants of Tourette's Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. *American Journal of Psychiatry*, **2019**, 176, 217-227	11.9	95
197	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. *Nature Neuroscience*, **2017**, 20, 1661-1668	25.5	95
196	A genome-wide association study of shared risk across psychiatric disorders implicates gene regulation during fetal neurodevelopment. *Nature Neuroscience*, **2019**, 22, 353-361	25.5	93
195	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, 1369-78	3.5	93
194	A Genetic Investigation of Sex Bias in the Prevalence of Attention-Deficit/Hyperactivity Disorder. *Biological Psychiatry*, **2018**, 83, 1044-1053	7.9	93
193	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, 15161-5	11.5	92
192	Common body mass index-associated variants confer risk of extreme obesity. *Human Molecular Genetics*, **2009**, 18, 3502-7	5.6	91
191	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. *Nature Communications*, **2018**, 9, 3391	17.4	90
190	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. *Neuron*, **2017**, 94, 486-499.e9	13.9	89
189	Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects. *Nature Communications*, **2018**, 9, 4038	17.4	87
188	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, 910-9	7.2	86
187	Mapping and characterization of structural variation in 17,795 human genomes. *Nature*, **2020**, 583, 83-89	50.4	84

186	A synthetic-diploid benchmark for accurate variant-calling evaluation. *Nature Methods*, **2018**, 15, 595-597	21.6	83
185	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	36.3	83
184	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	7.2	80
183	Genetic Differences in the Immediate Transcriptome Response to Stress Predict Risk-Related Brain Function and Psychiatric Disorders. *Neuron*, **2015**, 86, 1189-202	13.9	79
182	Reply to Cipriani et al. *European Journal of Human Genetics*, **2012**, 20, 3-3	5.3	78
181	Genome-wide association studies in an isolated founder population from the Pacific Island of Kosrae. *PLoS Genetics*, **2009**, 5, e1000365	6	75
180	Body dissatisfaction and drive for thinness in young adult twins. *International Journal of Eating Disorders*, **2005**, 37, 188-99	6.3	74
179	RICOPILI: Rapid Imputation for COnsortias PIpeLIne. *Bioinformatics*, **2020**, 36, 930-933	7.2	72
178	Autism spectrum disorder and attention deficit hyperactivity disorder have a similar burden of rare protein-truncating variants. *Nature Neuroscience*, **2019**, 22, 1961-1965	25.5	64
177	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. *Nature Neuroscience*, **2016**, 19, 1563-1565	25.5	63
176	Discovery of the first genome-wide significant risk loci for ADHD		62
175	The structure of perfectionism: a twin study. *Behavior Genetics*, **2004**, 34, 483-94	3.2	59
174	Joint Contributions of Rare Copy Number Variants and Common SNPs to Risk for Schizophrenia. *American Journal of Psychiatry*, **2019**, 176, 29-35	11.9	59
173	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. *American Journal of Human Genetics*, **2018**, 102, 1204-1211	11	59
172	Exome sequencing in amyotrophic lateral sclerosis implicates a novel gene, DNAJC7, encoding a heat-shock protein. *Nature Neuroscience*, **2019**, 22, 1966-1974	25.5	56
171	Estimation of Genetic Correlation via Linkage Disequilibrium Score Regression and Genomic Restricted Maximum Likelihood. *American Journal of Human Genetics*, **2018**, 102, 1185-1194	11	55
170	Spatiotemporal profile of postsynaptic interactomes integrates components of complex brain disorders. *Nature Neuroscience*, **2017**, 20, 1150-1161	25.5	54
169	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. *Nature Communications*, **2018**, 9, 2606	17.4	53

168	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	23.3	53
167	Genetic analysis of schizophrenia and bipolar disorder reveals polygenicity but also suggests new directions for molecular interrogation. *Current Opinion in Neurobiology*, **2015**, 30, 131-8	7.6	52
166	Exome sequencing in schizophrenia-affected parent-offspring trios reveals risk conferred by protein-coding de novo mutations. *Nature Neuroscience*, **2020**, 23, 185-193	25.5	52
165	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	10.6	51
164	Functional Architectures of Local and Distal Regulation of Gene Expression in Multiple Human Tissues. *American Journal of Human Genetics*, **2017**, 100, 605-616	11	50
163	Large-Scale trans-eQTLs Affect Hundreds of Transcripts and Mediate Patterns of Transcriptional Co-regulation. *American Journal of Human Genetics*, **2017**, 100, 581-591	11	46
162	Associations of CFHR1-CFHR3 deletion and a CFH SNP to age-related macular degeneration are not independent. *Nature Genetics*, **2010**, 42, 553-5; author reply 555-6	36.3	46
161	Phenotypic extremes in rare variant study designs. *European Journal of Human Genetics*, **2016**, 24, 924-30	5.3	43
160	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	6	43
159	New data and an old puzzle: the negative association between schizophrenia and rheumatoid arthritis. *International Journal of Epidemiology*, **2015**, 44, 1706-21	7.8	43
158	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. *Neuron*, **2018**, 98, 743-753.e4	13.9	42
157	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts. *Nature Genetics*, **2020**, 52, 634-639	36.3	41
156	A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. *Nature Communications*, **2016**, 7, 12342	17.4	41
155	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. *PLoS Genetics*, **2018**, 14, e1007329	6	41
154	Exome sequencing identifies rare coding variants in 10 genes which confer substantial risk for schizophrenia		39
153	Linkage to chromosome 1p36 for attention-deficit/hyperactivity disorder traits in school and home settings. *Biological Psychiatry*, **2008**, 64, 571-6	7.9	38
152	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, 437-52	11	37
151	Genetic Markers of ADHD-Related Variations in Intracranial Volume. *American Journal of Psychiatry*, **2019**, 176, 228-238	11.9	36

150	Genetic research in autism spectrum disorders. *Current Opinion in Pediatrics*, **2015**, 27, 685-91	3.2	36
149	Exome sequencing in suspected monogenic dyslipidemias. *Circulation: Cardiovascular Genetics*, **2015**, 8, 343-50		36
148	Mapping genomic loci implicates genes and synaptic biology in schizophrenia.. *Nature*, **2022**,	50.4	35
147	Haplotype Sharing Provides Insights into Fine-Scale Population History and Disease in Finland. *American Journal of Human Genetics*, **2018**, 102, 760-775	11	34
146	The positives, protocols, and perils of genome-wide association. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2008**, 147B, 1288-94	3.5	34
145	Intentional weight loss in young adults: sex-specific genetic and environmental effects. *Obesity*, **2005**, 13, 745-53		34
144	An open resource of structural variation for medical and population genetics		33
143	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	36.3	33
142	Genome-wide association study identifies 48 common genetic variants associated with handedness. *Nature Human Behaviour*, **2021**, 5, 59-70	12.8	33
141	Common risk variants identified in autism spectrum disorder		32
140	Inherited myeloproliferative neoplasm risk affects haematopoietic stem cells. *Nature*, **2020**, 586, 769-775	50.4	32
139	Discovery of rare variants for complex phenotypes. *Human Genetics*, **2016**, 135, 625-34	6.3	32
138	A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. *Diabetes*, **2017**, 66, 2019-2032	0.9	29
137	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, 573-80	11	28
136	Genome-wide association study identifies 30 Loci Associated with Bipolar Disorder		28
135	Evaluating the cardiovascular safety of sclerostin inhibition using evidence from meta-analysis of clinical trials and human genetics. *Science Translational Medicine*, **2020**, 12,	17.5	27
134	PhenoSpD: an integrated toolkit for phenotypic correlation estimation and multiple testing correction using GWAS summary statistics. *GigaScience*, **2018**, 7,	7.6	27
133	A note on the parameterization of Purcell's G x E model for ordinal and binary data. *Behavior Genetics*, **2009**, 39, 220-9	3.2	26

132	Attention-deficit/hyperactivity disorder and lifetime cannabis use: genetic overlap and causality. *Molecular Psychiatry*, **2020**, 25, 2493-2503	15.1	26
131	Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. *Nature Genetics*, **2021**, 53, 195-204	36.3	26
130	Quantifying the Genetic Correlation between Multiple Cancer Types. *Cancer Epidemiology Biomarkers and Prevention*, **2017**, 26, 1427-1435	4	25
129	Genetic profile for five common variants associated with age-related macular degeneration in densely affected families: a novel analytic approach. *European Journal of Human Genetics*, **2010**, 18, 496-501	5.3	25
128	Nonpaternity in linkage studies of extremely discordant sib pairs. *American Journal of Human Genetics*, **2002**, 70, 526-9	11	25
127	The genetics of neuropsychiatric diseases: looking in and beyond the exome. *Annual Review of Neuroscience*, **2015**, 38, 47-68	17	24
126	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. *Genome Research*, **2018**, 28, 968-974	9.7	23
125	Problems with Using Polygenic Scores to Select Embryos. *New England Journal of Medicine*, **2021**, 385, 78-86	59.2	23
124	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	6.8	22
123	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. *Nature Communications*, **2020**, 11, 3368	17.4	22
122	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179	8.2	22
121	Genome-wide association study of blood pressure response to methylphenidate treatment of attention-deficit/hyperactivity disorder. *Progress in Neuro-Psychopharmacology and Biological Psychiatry*, **2011**, 35, 466-72	5.5	21
120	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism		21
119	Genetic analyses identify widespread sex-differential participation bias. *Nature Genetics*, **2021**, 53, 663-671	36.3	20
118	A framework for the detection of de novo mutations in family-based sequencing data. *European Journal of Human Genetics*, **2017**, 25, 227-233	5.3	19
117	Signals of polygenic adaptation on height have been overestimated due to uncorrected population structure in genome-wide association studies		19
116	Disentangling polygenic associations between attention-deficit/hyperactivity disorder, educational attainment, literacy and language. *Translational Psychiatry*, **2019**, 9, 35	8.6	18
115	Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types		18

114	A global overview of pleiotropy and genetic architecture in complex traits		18
113	MTAG: Multi-Trait Analysis of GWAS		17
112	A Polygenic and Phenotypic Risk Prediction for Polycystic Ovary Syndrome Evaluated by Phenome-Wide Association Studies. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	17
111	A Comparison of Ten Polygenic Score Methods for Psychiatric Disorders Applied Across Multiple Cohorts. *Biological Psychiatry*, **2021**, 90, 611-620	7.9	17
110	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. *American Journal of Human Genetics*, **2020**, 107, 46-59	11	16
109	Population differences in the International Multi-Centre ADHD Gene Project. *Genetic Epidemiology*, **2008**, 32, 98-107	2.6	16
108	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. *Nature Medicine*, **2021**, 27, 1012-1024	50.5	16
107	Rare coding variants in ten genes confer substantial risk for schizophrenia.. *Nature*, **2022**,	50.4	16
106	Genetic variation associated with euphorigenic effects of d-amphetamine is associated with diminished risk for schizophrenia and attention deficit hyperactivity disorder. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 5968-73	11.5	15
105	No Reliable Association between Runs of Homozygosity and Schizophrenia in a Well-Powered Replication Study. *PLoS Genetics*, **2016**, 12, e1006343	6	15
104	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights		15
103	Genetic analyses identify widespread sex-differential participation bias		15
102	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. *International Journal of Epidemiology*, **2020**, 49, 1022-1031	7.8	15
101	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
100	Age at onset in sod1-mediated amyotrophic lateral sclerosis shows familiality. *Neurogenetics*, **2007**, 8, 235-6	3	14
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35	Bootstrat: Population Informed Bootstrapping for Rare Variant Tests		2
34	Insights into the genetic epidemiology of Crohn and rare diseases in the Ashkenazi Jewish population		2
33	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations		2
32	Tractor: A framework allowing for improved inclusion of admixed individuals in large-scale association studies		2
31	Ancestry May Confound Genetic Machine Learning: Candidate-Gene Prediction of Opioid Use Disorder as an Example		2
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