Benjamin M Neale

List of Publications by Year in Descending Order

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	Sex-Dependent Shared and Nonshared Genetic Architecture Across Mood and Psychotic Disorders. *Biological Psychiatry*, **2022**, 91, 102-117	7.9	11
292	Mapping genomic loci implicates genes and synaptic biology in schizophrenia.. *Nature*, **2022**,	50.4	35
291	Rare coding variants in ten genes confer substantial risk for schizophrenia.. *Nature*, **2022**,	50.4	16
290	Getting genetic ancestry right for science and society.. *Science*, **2022**, 376, 250-252	33.3	2
289	Challenges and Opportunities for Developing More Generalizable Polygenic Risk Scores.. *Annual Review of Biomedical Data Science*, **2022**,	5.6	1
288	GWAS significance thresholds for deep phenotyping studies can depend upon minor allele frequencies and sample size. *Molecular Psychiatry*, **2021**, 26, 2048-2055	15.1	7
287	Reply to: On powerful GWAS in admixed populations. *Nature Genetics*, **2021**, 53, 1634-1635	36.3	0
286	Response to comment on "Evaluating the cardiovascular safety of sclerostin inhibition using evidence from meta-analysis of clinical trials and human genetics". *Science Translational Medicine*, **2021**, 13, eabf4530	17.5	
285	Ancestry may confound genetic machine learning: Candidate-gene prediction of opioid use disorder as an example. *Drug and Alcohol Dependence*, **2021**, 229, 109115	4.9	0
284	Response to Comment on "Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior". *Science*, **2021**, 371,	33.3	2
283	Genetic analyses identify widespread sex-differential participation bias. *Nature Genetics*, **2021**, 53, 663-671	36.3	20
282	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. *American Journal of Human Genetics*, **2021**, 108, 656-668	11	10
281	A Comparison of Ten Polygenic Score Methods for Psychiatric Disorders Applied Across Multiple Cohorts. *Biological Psychiatry*, **2021**, 90, 611-620	7.9	17
280	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
279	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. *Epilepsia*, **2021**, 62, 1518-1527	6.4	1
278	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	11	6
277	Examining Sex-Differentiated Genetic Effects Across Neuropsychiatric and Behavioral Traits. *Biological Psychiatry*, **2021**, 89, 1127-1137	7.9	12

276	Hematopoietic mosaic chromosomal alterations increase the risk for diverse types of infection. *Nature Medicine*, **2021**, 27, 1012-1024	50.5	16
275	Genome-wide association study identifies 48 common genetic variants associated with handedness. *Nature Human Behaviour*, **2021**, 5, 59-70	12.8	33
274	Synaptic processes and immune-related pathways implicated in Tourette syndrome. *Translational Psychiatry*, **2021**, 11, 56	8.6	11
273	Risk variants and polygenic architecture of disruptive behavior disorders in the context of attention-deficit/hyperactivity disorder. *Nature Communications*, **2021**, 12, 576	17.4	3
272	Estimating heritability and its enrichment in tissue-specific gene sets in admixed populations. *Human Molecular Genetics*, **2021**, 30, 1521-1534	5.6	6
271	Genome-wide association study of pediatric obsessive-compulsive traits: shared genetic risk between traits and disorder. *Translational Psychiatry*, **2021**, 11, 91	8.6	10
270	Mapping the human genetic architecture of COVID-19. *Nature*, **2021**,	50.4	162
269	Problems with Using Polygenic Scores to Select Embryos. *New England Journal of Medicine*, **2021**, 385, 78-86	59.2	23
268	Clinical Conditions and Their Impact on Utility of Genetic Scores for Prediction of Acute Coronary Syndrome. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003283	5.2	2
267	Addendum: The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2021**, 597, E3-E4	50.4	3
266	Investigating Shared Genetic Basis Across Tourette Syndrome and Comorbid Neurodevelopmental Disorders Along the Impulsivity-Compulsivity Spectrum. *Biological Psychiatry*, **2021**, 90, 317-327	7.9	12
265	Human genetic analyses of organelles highlight the nucleus in age-related trait heritability. *ELife*, **2021**, 10,	8.9	1
264	Elevated common variant genetic risk for tourette syndrome in a densely-affected pedigree. *Molecular Psychiatry*, **2021**,	15.1	1
263	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
262	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. *Molecular Psychiatry*, **2021**, 26, 5239-5250	15.1	3
261	Habitual sleep disturbances and migraine: a Mendelian randomization study. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2370-2380	5.3	6
260	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts. *Nature Genetics*, **2020**, 52, 634-639	36.3	41
259	Non-parametric Polygenic Risk Prediction via Partitioned GWAS Summary Statistics. *American Journal of Human Genetics*, **2020**, 107, 46-59	11	16

258	The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature*, **2020**, 581, 434-443	50.4	2278
257	A structural variation reference for medical and population genetics. *Nature*, **2020**, 581, 444-451	50.4	223
256	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
255	Mapping and characterization of structural variation in 17,795 human genomes. *Nature*, **2020**, 583, 83-89	50.4	84
254	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. *Nature Communications*, **2020**, 11, 3368	17.4	22
253	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
252	Statistical Power and the Classical Twin Design. *Twin Research and Human Genetics*, **2020**, 23, 87-89	2.2	0
251	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
250	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
249	Hematopoietic mosaic chromosomal alterations and risk for infection among 767,891 individuals without blood cancer **2020**,		2
248	Hematopoietic mosaic chromosomal alterations and risk for infection among 767,891 individuals without blood cancer **2020**,		5
247	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
246	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
245	Inherited myeloproliferative neoplasm risk affects haematopoietic stem cells. *Nature*, **2020**, 586, 769-775	50.4	32
244	The genetic architecture of sporadic and multiple consecutive miscarriage. *Nature Communications*, **2020**, 11, 5980	17.4	11
243	A data-driven medication score predicts 10-year mortality among aging adults. *Scientific Reports*, **2020**, 10, 15760	4.9	1
242	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
241	Attention-deficit/hyperactivity disorder and lifetime cannabis use: genetic overlap and causality. *Molecular Psychiatry*, **2020**, 25, 2493-2503	15.1	26

240	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
239	RICOPILI: Rapid Imputation for COnsortias PIpeLIne. *Bioinformatics*, **2020**, 36, 930-933	7.2	72
238	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
237	Large-scale GWAS reveals insights into the genetic architecture of same-sex sexual behavior. *Science*, **2019**, 365,	33.3	139
236	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
235	Disentangling polygenic associations between attention-deficit/hyperactivity disorder, educational attainment, literacy and language. *Translational Psychiatry*, **2019**, 9, 35	8.6	18
234	Estimation of metabolic syndrome heritability in three large populations including full pedigree and genomic information. *Human Genetics*, **2019**, 138, 739-748	6.3	2
233	Polygenic adaptation on height is overestimated due to uncorrected stratification in genome-wide association studies. *ELife*, **2019**, 8,	8.9	166
232	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
231	Genome-wide association study identifies 30 loci associated with bipolar disorder. *Nature Genetics*, **2019**, 51, 793-803	36.3	662
230	Genetic Markers of ADHD-Related Variations in Intracranial Volume. *American Journal of Psychiatry*, **2019**, 176, 228-238	11.9	36
229	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
228	Clinical use of current polygenic risk scores may exacerbate health disparities. *Nature Genetics*, **2019**, 51, 584-591	36.3	711
227	The Manifestation Of Genetic Risk For Attention Deficit Hyperactivity Disorder In Females And Males In The General Population. *European Neuropsychopharmacology*, **2019**, 29, S723-S724	1.2	
226	Population-based identity-by-descent mapping combined with exome sequencing to detect rare risk variants for schizophrenia. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2019**, 180, 223-231	3.5	2
225	Advanced Paternal Age and Early Onset of Schizophrenia in Sporadic Cases: Not Confounded by Parental Polygenic Risk for Schizophrenia. *Biological Psychiatry*, **2019**, 86, 56-64	7.9	10
224	Identification of common genetic risk variants for autism spectrum disorder. *Nature Genetics*, **2019**, 51, 431-444	36.3	746
223	A global overview of pleiotropy and genetic architecture in complex traits. *Nature Genetics*, **2019**, 51, 1339-1348	36.3	311

222	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
221	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
220	Recent advances in understanding of attention deficit hyperactivity disorder (ADHD): how genetics are shaping our conceptualization of this disorder. *F1000Research*, **2019**, 8,	3.6	4
219	Comparative genetic architectures of schizophrenia in East Asian and European populations. *Nature Genetics*, **2019**, 51, 1670-1678	36.3	185
218	Genome studies must account for history-Response. *Science*, **2019**, 366, 1461-1462	33.3	4
217	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
216	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
215	GWAS Identifies Risk Locus for Erectile Dysfunction and Implicates Hypothalamic Neurobiology and Diabetes in Etiology. *American Journal of Human Genetics*, **2019**, 104, 157-163	11	12
214	Predicting Polygenic Risk of Psychiatric Disorders. *Biological Psychiatry*, **2019**, 86, 97-109	7.9	170
213	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
212	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. *Nature Genetics*, **2019**, 51, 63-75	36.3	826
211	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
210	Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types. *Nature Genetics*, **2018**, 50, 621-629	36.3	400
209	Transcriptome-wide association study of schizophrenia and chromatin activity yields mechanistic disease insights. *Nature Genetics*, **2018**, 50, 538-548	36.3	222
208	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. *Science*, **2018**, 359, 693-697	33.3	547
207	Multi-trait analysis of genome-wide association summary statistics using MTAG. *Nature Genetics*, **2018**, 50, 229-237	36.3	339
206	Common Variant Burden Contributes to the Familial Aggregation of Migraine in 1,589 Families. *Neuron*, **2018**, 98, 743-753.e4	13.9	42
205	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

204	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
203	Haplotype Sharing Provides Insights into Fine-Scale Population History and Disease in Finland. *American Journal of Human Genetics*, **2018**, 102, 760-775	11	34
202	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
201	Deep coverage whole genome sequences and plasma lipoprotein(a) in individuals of European and African ancestries. *Nature Communications*, **2018**, 9, 2606	17.4	53
200	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. *Nature Communications*, **2018**, 9, 3391	17.4	90
199	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
198	Insights into the genetic epidemiology of Crohn's and rare diseases in the Ashkenazi Jewish population. *PLoS Genetics*, **2018**, 14, e1007329	6	41
197	A synthetic-diploid benchmark for accurate variant-calling evaluation. *Nature Methods*, **2018**, 15, 595-597	21.6	83
196	A Genetic Investigation of Sex Bias in the Prevalence of Attention-Deficit/Hyperactivity Disorder. *Biological Psychiatry*, **2018**, 83, 1044-1053	7.9	93
195	Transancestral GWAS of alcohol dependence reveals common genetic underpinnings with psychiatric disorders. *Nature Neuroscience*, **2018**, 21, 1656-1669	25.5	257
194	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
193	Genome-wide de novo risk score implicates promoter variation in autism spectrum disorder. *Science* , **2018**, 362,	33.3	134
192	Genetics of blood lipids among ~300,000 multi-ethnic participants of the Million Veteran Program. *Nature Genetics*, **2018**, 50, 1514-1523	36.3	260
191	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
190	Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects. *Nature Communications*, **2018**, 9, 4038	17.4	87
189	Large-scale meta-analysis highlights the hypothalamic-pituitary-gonadal axis in the genetic regulation of menstrual cycle length. *Human Molecular Genetics*, **2018**, 27, 4323-4332	5.6	11
188	PhenoSpD: an integrated toolkit for phenotypic correlation estimation and multiple testing correction using GWAS summary statistics. *GigaScience*, **2018**, 7,	7.6	27
187	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

186	Base-specific mutational intolerance near splice sites clarifies the role of nonessential splice nucleotides. *Genome Research*, **2018**, 28, 968-974	9.7	23
185	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
184	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
183	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
182	De Novo Coding Variants Are Strongly Associated with Tourette Disorder. *Neuron*, **2017**, 94, 486-499.e9	13.9	89
181	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. *Neuron*, **2017**, 94, 1101-1111.e7	13.9	103
180	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
179	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
178	Human Demographic History Impacts Genetic Risk Prediction across Diverse Populations. *American Journal of Human Genetics*, **2017**, 100, 635-649	11	665
177	Linkage disequilibrium-dependent architecture of human complex traits shows action of negative selection. *Nature Genetics*, **2017**, 49, 1421-1427	36.3	204
176	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. *Nature Neuroscience*, **2017**, 20, 1661-1668	25.5	95
175	Quantifying the Genetic Correlation between Multiple Cancer Types. *Cancer Epidemiology Biomarkers and Prevention*, **2017**, 26, 1427-1435	4	25
174	Spatiotemporal profile of postsynaptic interactomes integrates components of complex brain disorders. *Nature Neuroscience*, **2017**, 20, 1150-1161	25.5	54
173	223. Genome-Wide Association Study of Posttraumatic Stress Disorder Symptom Domains in Two Cohorts of United States Army Soldiers. *Biological Psychiatry*, **2017**, 81, S91-S92	7.9	2
172	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
171	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
170	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
169	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. *Scientific Data*, **2017**, 4, 170179	8.2	22

168	Phenome-wide heritability analysis of the UK Biobank. *PLoS Genetics*, **2017**, 13, e1006711	6	131
167	Phenotypic extremes in rare variant study designs. *European Journal of Human Genetics*, **2016**, 24, 924-30	5.3	43
166	Analysis of protein-coding genetic variation in 60,706 humans. *Nature*, **2016**, 536, 285-91	50.4	6940
165	A protein-truncating R179X variant in RNF186 confers protection against ulcerative colitis. *Nature Communications*, **2016**, 7, 12342	17.4	41
164	Association of Exome Sequences With Cardiovascular Traits Among Blacks in the Jackson Heart Study. *Circulation: Cardiovascular Genetics*, **2016**, 9, 368-74		7
163	Association Strategies **2016**, 133-139		
162	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. *Nature Genetics*, **2016**, 48, 856-66	36.3	355
161	Genetic Effect of Chemotherapy Exposure in Children of Testicular Cancer Survivors. *Clinical Cancer Research*, **2016**, 22, 2183-9	12.9	10
160	Genetic risk for autism spectrum disorders and neuropsychiatric variation in the general population. *Nature Genetics*, **2016**, 48, 552-5	36.3	238
159	Genetic influences on schizophrenia and subcortical brain volumes: large-scale proof of concept. *Nature Neuroscience*, **2016**, 19, 420-431	25.5	163
158	No Reliable Association between Runs of Homozygosity and Schizophrenia in a Well-Powered Replication Study. *PLoS Genetics*, **2016**, 12, e1006343	6	15
157	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
156	Discovery of rare variants for complex phenotypes. *Human Genetics*, **2016**, 135, 625-34	6.3	32
155	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. *American Journal of Human Genetics*, **2016**, 98, 857-868	11	14
154	The statistical properties of gene-set analysis. *Nature Reviews Genetics*, **2016**, 17, 353-64	30.1	162
153	Genome-wide Association Studies of Posttraumatic Stress Disorder in 2 Cohorts of US Army Soldiers. *JAMA Psychiatry*, **2016**, 73, 695-704	14.5	114
152	Ultra-rare disruptive and damaging mutations influence educational attainment in the general population. *Nature Neuroscience*, **2016**, 19, 1563-1565	25.5	63
151	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

150	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
149	The genetics of neuropsychiatric diseases: looking in and beyond the exome. *Annual Review of Neuroscience*, **2015**, 38, 47-68	17	24
148	Modeling Linkage Disequilibrium Increases Accuracy of Polygenic Risk Scores. *American Journal of Human Genetics*, **2015**, 97, 576-92	11	649
147	Partitioning heritability by functional annotation using genome-wide association summary statistics. *Nature Genetics*, **2015**, 47, 1228-35	36.3	1143
146	An atlas of genetic correlations across human diseases and traits. *Nature Genetics*, **2015**, 47, 1236-41	36.3	1841
145	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
144	Contrasting genetic architectures of schizophrenia and other complex diseases using fast variance-components analysis. *Nature Genetics*, **2015**, 47, 1385-92	36.3	299
143	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
142	Genetic research in autism spectrum disorders. *Current Opinion in Pediatrics*, **2015**, 27, 685-91	3.2	36
141	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
140	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
139	Exome sequencing in suspected monogenic dyslipidemias. *Circulation: Cardiovascular Genetics*, **2015**, 8, 343-50		36
138	LD Score regression distinguishes confounding from polygenicity in genome-wide association studies. *Nature Genetics*, **2015**, 47, 291-5	36.3	2096
137	Efficient Bayesian mixed-model analysis increases association power in large cohorts. *Nature Genetics*, **2015**, 47, 284-90	36.3	758
136	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
135	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
134	Whole-genome analyses of whole-brain data: working within an expanded search space. *Nature Neuroscience*, **2014**, 17, 791-800	25.5	98
133	Partitioning heritability of regulatory and cell-type-specific variants across 11 common diseases. *American Journal of Human Genetics*, **2014**, 95, 535-52	11	411

132	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
131	A framework for the interpretation of de novo mutation in human disease. *Nature Genetics*, **2014**, 46, 944-50	36.3	656
130	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
129	Rare deleterious mutations of the gene EFR3A in autism spectrum disorders. *Molecular Autism*, **2014**, 5, 31	6.5	11
128	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
127	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
126	Clozapine-induced agranulocytosis is associated with rare HLA-DQB1 and HLA-B alleles. *Nature Communications*, **2014**, 5, 4757	17.4	118
125	Clonal hematopoiesis and blood-cancer risk inferred from blood DNA sequence. *New England Journal of Medicine*, **2014**, 371, 2477-87	59.2	1855
124	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
123	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. *Nature Genetics*, **2013**, 45, 1150-9	36.3	1153
122	Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nature Genetics*, **2013**, 45, 1345-52	36.3	597
121	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
120	Exome array analysis identifies new loci and low-frequency variants influencing insulin processing and secretion. *Nature Genetics*, **2013**, 45, 197-201	36.3	212
119	Rare complete knockouts in humans: population distribution and significant role in autism spectrum disorders. *Neuron*, **2013**, 77, 235-42	13.9	190
118	A mega-analysis of genome-wide association studies for major depressive disorder. *Molecular Psychiatry*, **2013**, 18, 497-511	15.1	853
117	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
116	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
115	Genome-wide meta-analysis identifies new susceptibility loci for migraine. *Nature Genetics*, **2013**, 45, 912-917	36.3	276

114	High loading of polygenic risk for ADHD in children with comorbid aggression. *American Journal of Psychiatry*, **2013**, 170, 909-16	11.9	110
113	Analysis of rare, exonic variation amongst subjects with autism spectrum disorders and population controls. *PLoS Genetics*, **2013**, 9, e1003443	6	108
112	Partitioning the heritability of Tourette syndrome and obsessive compulsive disorder reveals differences in genetic architecture. *PLoS Genetics*, **2013**, 9, e1003864	6	189
111	Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell*, **2012**, 149, 525-37	56.2	441
110	Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature*, **2012**, 485, 242-5	50.4	1300
109	Extremely low-coverage sequencing and imputation increases power for genome-wide association studies. *Nature Genetics*, **2012**, 44, 631-5	36.3	184
108	Exome sequencing and the genetic basis of complex traits. *Nature Genetics*, **2012**, 44, 623-30	36.3	303
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64	Multi-ancestry meta-analysis of asthma identifies novel associations and highlights the value of increased power and diversity		2
63	Haplotype sharing provides insights into fine-scale population history and disease in Finland		2
62	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals		4
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60	Genetic predisposition to myeloproliferative neoplasms implicates hematopoietic stem cell biology	1
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58	Subtle stratification confounds estimates of heritability from rare variants	14
57	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	2
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46	A genetic investigation of sex bias in the prevalence of attention deficit hyperactivity disorder	3
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38	Ancestry May Confound Genetic Machine Learning: Candidate-Gene Prediction of Opioid Use Disorder as an Example	2
37	Exome sequencing identifies rare coding variants in 10 genes which confer substantial risk for schizophrenia	39
36	An efficient and accurate frailty model approach for genome-wide survival association analysis controlling for population structure and relatedness in large-scale biobanks	5
35	New synthetic-diploid benchmark for accurate variant calling evaluation	8
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32	Trans-ancestral GWAS of alcohol dependence reveals common genetic underpinnings with psychiatric disorders	7
31	ASD and ADHD have a similar burden of rare protein-truncating variants	12
30	Enrichment of rare protein truncating variants in amyotrophic lateral sclerosis patients	4
29	Signals of polygenic adaptation on height have been overestimated due to uncorrected population structure in genome-wide association studies	19
28	Non-parametric polygenic risk prediction using partitioned GWAS summary statistics	3
27	Comparative genetic architectures of schizophrenia in East Asian and European populations	8
26	Large-scale exome sequencing study implicates both developmental and functional changes in the neurobiology of autism	21
25	Schizophrenia risk conferred by protein-coding de novo mutations	4

24	A global overview of pleiotropy and genetic architecture in complex traits	18
23	Estimating heritability and its enrichment in tissue-specific gene sets in admixed populations	4
22	Mapping and characterization of structural variation in 17,795 deeply sequenced human genomes	12
21	Genome wide meta-analysis identifies genomic relationships, novel loci, and pleiotropic mechanisms across eight psychiatric disorders	10
20	The mutational constraint spectrum quantified from variation in 141,456 humans	381
19	The genetic architecture of sporadic and recurrent miscarriage	2
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17	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts	7
16	RICOPILI: Rapid Imputation for COnsortias PIpeLIne	8
15	Polygenic and clinical risk scores and their impact on age at onset of cardiometabolic diseases and common cancers	6
14	Cross-disorder GWAS meta-analysis for Attention Deficit/Hyperactivity Disorder, Autism Spectrum Disorder, Obsessive Compulsive Disorder, and Tourette Syndrome	2
13	Identification of risk variants and characterization of the polygenic architecture of disruptive behavior disorders in the context of ADHD	1
12	Functional partitioning of local and distal gene expression regulation in multiple human tissues	2
11	Base-specific mutational intolerance near splice-sites clarifies role of non-essential splice nucleotides	1
10	Quantifying the impact of rare and ultra-rare coding variation across the phenotypic spectrum	1
9	Multi-Ancestry Meta-Analysis yields novel genetic discoveries and ancestry-specific associations	3
8	Incorporating family history of disease improves polygenic risk scores in diverse populations	4
7	Systematic single-variant and gene-based association testing of 3,700 phenotypes in 281,850 UK Biobank exomes	6

6	Human genetic analyses of organelles highlight the nucleus in age-related trait heritability	1
5	A cross-disorder dosage sensitivity map of the human genome	5
4	Set-based rare variant association tests for biobank scale sequencing data sets	1
3	Analysis of genetic dominance in the UK Biobank	1
2	Differences in the genetic architecture of common and rare variants in childhood, persistent and late-diagnosed attention deficit hyperactivity disorder	1
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