Goncalo Abecasis

List of Publications by Year
in descending order

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380	222,063	167 155	55 391
papers	citations	h-index	g-index

424	424	424	181587
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The Sequence Alignment/Map format and SAMtools. Bioinformatics, 2009, 25, 2078-2079.	1.8	49,124
2	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
3	The variant call format and VCFtools. Bioinformatics, 2011, 27, 2156-2158.	1.8	11,326
4	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
5	An integrated map of genetic variation from 1,092 human genomes. Nature, 2012, 491, 56-65.	13.7	7,199
6	A second generation human haplotype map of over 3.1 million SNPs. Nature, 2007, 449, 851-861.	13.7	4,137
7	METAL: fast and efficient meta-analysis of genomewide association scans. Bioinformatics, 2010, 26, 2190-2191.	1.8	4,046
8	Genetic studies of body mass index yield new insights for obesity biology. Nature, 2015, 518, 197-206.	13.7	3,823
9	Biological, clinical and population relevance of 95 loci for blood lipids. Nature, 2010, 466, 707-713.	13.7	3,249
10	Merlinâ€"rapid analysis of dense genetic maps using sparse gene flow trees. Nature Genetics, 2002, 30, 97-101.	9.4	3,100
11	Next-generation genotype imputation service and methods. Nature Genetics, 2016, 48, 1284-1287.	9.4	2,828
12	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. Nature Genetics, 2010, 42, 937-948.	9.4	2,634
13	A Genome-Wide Association Study of Type 2 Diabetes in Finns Detects Multiple Susceptibility Variants. Science, 2007, 316, 1341-1345.	6.0	2,534
14	Genome-wide association studies for complex traits: consensus, uncertainty and challenges. Nature Reviews Genetics, 2008, 9, 356-369.	7.7	2,496
15	A reference panel of 64,976 haplotypes for genotype imputation. Nature Genetics, 2016, 48, 1279-1283.	9.4	2,421
16	LocusZoom: regional visualization of genome-wide association scan results. Bioinformatics, 2010, 26, 2336-2337.	1.8	2,349
17	Plasma HDL cholesterol and risk of myocardial infarction: a mendelian randomisation study. Lancet, The, 2012, 380, 572-580.	6.3	1,937
18	Defining the role of common variation in the genomic and biological architecture of adult human height. Nature Genetics, 2014, 46, 1173-1186.	9.4	1,818

#	Article	IF	Citations
19	Hundreds of variants clustered in genomic loci and biological pathways affect human height. Nature, 2010, 467, 832-838.	13.7	1,789
20	Genome-wide detection and characterization of positive selection in human populations. Nature, 2007, 449, 913-918.	13.7	1,788
21	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. Nature Genetics, 2012, 44, 981-990.	9.4	1,748
22	MaCH: using sequence and genotype data to estimate haplotypes and unobserved genotypes. Genetic Epidemiology, 2010, 34, 816-834.	0.6	1,718
23	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. Nature Genetics, 2010, 42, 579-589.	9.4	1,631
24	Fast and accurate genotype imputation in genome-wide association studies through pre-phasing. Nature Genetics, 2012, 44, 955-959.	9.4	1,592
25	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. Nature Genetics, 2009, 41, 25-34.	9.4	1,572
26	Evolution and Functional Impact of Rare Coding Variation from Deep Sequencing of Human Exomes. Science, 2012, 337, 64-69.	6.0	1,535
27	Replicating genotypeâ€“phenotype associations. Nature, 2007, 447, 655-660.	13.7	1,509
28	Newly identified loci that influence lipid concentrations and risk of coronary artery disease. Nature Genetics, 2008, 40, 161-169.	9.4	1,488
29	Genome-Wide Association Scan Shows Genetic Variants in the FTO Gene Are Associated with Obesity-Related Traits. PLoS Genetics, 2007, 3, e115.	1.5	1,446
30	Genetic variants regulating ORMDL3 expression contribute to the risk of childhood asthma. Nature, 2007, 448, 470-473.	13.7	1,446
31	Reference-based phasing using the Haplotype Reference Consortium panel. Nature Genetics, 2016, 48, 1443-1448.	9.4	1,357
32	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. Nature Genetics, 2018, 50, 1505-1513.	9.4	1,331
33	New genetic loci link adipose and insulin biology to body fat distribution. Nature, 2015, 518, 187-196.	13.7	1,328
34	Association studies of up to 1.2 million individuals yield new insights into the genetic etiology of tobacco and alcohol use. Nature Genetics, 2019, 51, 237-244.	9.4	1,307
35	Common variants at 30 loci contribute to polygenic dyslipidemia. Nature Genetics, 2009, 41, 56-65.	9.4	1,234
36	A Note on Exact Tests of Hardy-Weinberg Equilibrium. American Journal of Human Genetics, 2005, 76, 887-893.	2.6	1,232

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37	Genome-wide scan reveals association of psoriasis with IL-23 and NF-Î²B pathways. Nature Genetics, 2009, 41, 199-204.	9.4	1,229
38	Common variants near MC4R are associated with fat mass, weight and risk of obesity. Nature Genetics, 2008, 40, 768-775.	9.4	1,179
39	A large genome-wide association study of age-related macular degeneration highlights contributions of rare and common variants. Nature Genetics, 2016, 48, 134-143.	9.4	1,167
40	Joint analysis is more efficient than replication-based analysis for two-stage genome-wide association studies. Nature Genetics, 2006, 38, 209-213.	9.4	1,166
41	Genome-wide association study identifies eight loci associated with blood pressure. Nature Genetics, 2009, 41, 666-676.	9.4	1,104
42	Sequencing of 53,831 diverse genomes from the NHLBI TOPMed Program. Nature, 2021, 590, 290-299.	13.7	1,069
43	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. Nature Genetics, 2014, 46, 234-244.	9.4	959
44	The genetic architecture of type 2 diabetes. Nature, 2016, 536, 41-47.	13.7	952
45	Loss-of-Function Mutations inAPOC3,Triglycerides, and Coronary Disease. New England Journal of Medicine, 2014, 371, 22-31.	13.9	936
46	Genetic analysis of over 1 million people identifies 535 new loci associated with blood pressure traits. Nature Genetics, 2018, 50, 1412-1425.	9.4	924
47	Genotype Imputation. Annual Review of Genomics and Human Genetics, 2009, 10, 387-406.	2.5	920
48	Analysis of 6,515 exomes reveals the recent origin of most human protein-coding variants. Nature, 2013, 493, 216-220.	13.7	898
49	Efficiently controlling for case-control imbalance and sample relatedness in large-scale genetic association studies. Nature Genetics, 2018, 50, 1335-1341.	9.4	896
50	A genome-wide association study of global gene expression. Nature Genetics, 2007, 39, 1202-1207.	9.4	882
51	Identification of 15 new psoriasis susceptibility loci highlights the role of innate immunity. Nature Genetics, 2012, 44, 1341-1348.	9.4	848
52	Rare-Variant Association Analysis: Study Designs and Statistical Tests. American Journal of Human Genetics, 2014, 95, 5-23.	2.6	837
53	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. Nature Genetics, 2010, 42, 949-960.	9.4	836
54	Mapping complex disease traits with global gene expression. Nature Reviews Genetics, 2009, 10, 184-194.	7.7	790

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55	Common variants associated with plasma triglycerides and risk for coronary artery disease. Nature Genetics, 2013, 45, 1345-1352.	9.4	754
56	Seven new loci associated with age-related macular degeneration. Nature Genetics, 2013, 45, 433-439.	9.4	687
57	Variants in MTNR1B influence fasting glucose levels. Nature Genetics, 2009, 41, 77-81.	9.4	662
58	An Abundance of Rare Functional Variants in 202 Drug Target Genes Sequenced in 14,002 People. Science, 2012, 337, 100-104.	6.0	626
59	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. Diabetes, 2017, 66, 2888-2902.	0.3	615
60	Meta-analysis and imputation refines the association of 15q25 with smoking quantity. Nature Genetics, 2010, 42, 436-440.	9.4	581
61	Exome sequencing identifies rare LDLR and APOA5 alleles conferring risk for myocardial infarction. Nature, 2015, 518, 102-106.	13.7	581
62	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
63	Genome-wide association study shows BCL11A associated with persistent fetal hemoglobin and amelioration of the phenotype of Î²-thalassemia. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 1620-1625.	3.3	561
64	Biobank-driven genomic discovery yields new insight into atrial fibrillation biology. Nature Genetics, 2018, 50, 1234-1239.	9.4	547
65	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	13.7	544
66	Identification of ten loci associated with height highlights new biological pathways in human growth. Nature Genetics, 2008, 40, 584-591.	9.4	537
67	Sequence and Haplotype Analysis Supports HLA-C as the Psoriasis Susceptibility 1 Gene. American Journal of Human Genetics, 2006, 78, 827-851.	2.6	529
68	Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. Nature Genetics, 2011, 43, 1131-1138.	9.4	501
69	Genetics of blood lipids among ~300,000 multi-ethnic participants of the Million Veteran Program. Nature Genetics, 2018, 50, 1514-1523.	9.4	497
70	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	9.4	482
71	Genome-wide association study in individuals of South Asian ancestry identifies six new type 2 diabetes susceptibility loci. Nature Genetics, 2011, 43, 984-989.	9.4	481
72	Genetic variants near TIMP3 and high-density lipoproteinâ€“associated loci influence susceptibility to age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 7401-7406.	3.3	475

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73	Exome-wide association study of plasma lipids in >300,000 individuals. Nature Genetics, 2017, 49, 1758-1766.	9.4	470
74	Heritability of Cardiovascular and Personality Traits in 6,148 Sardinians. PLoS Genetics, 2006, 2, e132.	1.5	468
75	Computationally efficient whole-genome regression for quantitative and binary traits. Nature Genetics, 2021, 53, 1097-1103.	9.4	457
76	Genome-Wide Association Scan Meta-Analysis Identifies Three Loci Influencing Adiposity and Fat Distribution. PLoS Genetics, 2009, 5, e1000508.	1.5	453
77	The Metabochip, a Custom Genotyping Array for Genetic Studies of Metabolic, Cardiovascular, and Anthropometric Traits. PLoS Genetics, 2012, 8, e1002793.	1.5	448
78	Detecting and Estimating Contamination of Human DNA Samples in Sequencing and Array-Based Genotype Data. American Journal of Human Genetics, 2012, 91, 839-848.	2.6	441
79	Rare variant in scavenger receptor BI raises HDL cholesterol and increases risk of coronary heart disease. Science, 2016, 351, 1166-1171.	6.0	438
80	Shared genetic origin of asthma, hay fever and eczema elucidates allergic disease biology. Nature Genetics, 2017, 49, 1752-1757.	9.4	432
81	Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. Nature Genetics, 2011, 43, 1005-1011.	9.4	403
82	Genome-wide association study of PR interval. Nature Genetics, 2010, 42, 153-159.	9.4	400
83	A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 16227-16232.	3.3	398
84	Common Variants at 10 Genomic Loci Influence Hemoglobin A1C Levels via Glycemic and Nonglycemic Pathways. Diabetes, 2010, 59, 3229-3239.	0.3	387
85	Inactivating Mutations in <i>NPC1L1</i> and Protection from Coronary Heart Disease. New England Journal of Medicine, 2014, 371, 2072-2082.	13.9	386
86	Family-Based Association Tests for Genomewide Association Scans. American Journal of Human Genetics, 2007, 81, 913-926.	2.6	383
87	FTO genotype is associated with phenotypic variability of body mass index. Nature, 2012, 490, 267-272.	13.7	383
88	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
89	Exome sequencing and analysis of 454,787 UK Biobank participants. Nature, 2021, 599, 628-634.	13.7	377
90	Gene polymorphism in Netherton and common atopic disease. Nature Genetics, 2001, 29, 175-178.	9.4	376

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91	A first-generation linkage disequilibrium map of human chromosome 22. Nature, 2002, 418, 544-548.	13.7	376
92	Inherited causes of clonal haematopoiesis in 97,691 whole genomes. Nature, 2020, 586, 763-768.	13.7	376
93	PEDSTATS: descriptive statistics, graphics and quality assessment for gene mapping data. Bioinformatics, 2005, 21, 3445-3447.	1.8	374
94	Sex-stratified Genome-wide Association Studies Including 270,000 Individuals Show Sexual Dimorphism in Genetic Loci for Anthropometric Traits. PLoS Genetics, 2013, 9, e1003500.	1.5	371
95	Common variants in the GDF5-UQCC region are associated with variation in human height. Nature Genetics, 2008, 40, 198-203.	9.4	369
96	Genetic Variants Influencing Circulating Lipid Levels and Risk of Coronary Artery Disease. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 2264-2276.	1.1	369
97	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. Nature, 2020, 586, 749-756.	13.7	369
98	RVTESTS: an efficient and comprehensive tool for rare variant association analysis using sequence data. Bioinformatics, 2016, 32, 1423-1426.	1.8	366
99	Using haplotype blocks to map human complex trait loci. Trends in Genetics, 2003, 19, 135-140.	2.9	360
100	Unified representation of genetic variants. Bioinformatics, 2015, 31, 2202-2204.	1.8	357
101	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
102	Common variants at ten loci modulate the QT interval duration in the QTSCD Study. Nature Genetics, 2009, 41, 407-414.	9.4	356
103	Genetic linkage of childhood atopic dermatitis to psoriasis susceptibility loci. Nature Genetics, 2001, 27, 372-373.	9.4	353
104	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	13.7	353
105	Genome-Wide Association Study of Plasma Polyunsaturated Fatty Acids in the InCHIANTI Study. PLoS Genetics, 2009, 5, e1000338.	1.5	351
106	Impact of common genetic determinants of Hemoglobin A1c on type 2 diabetes risk and diagnosis in ancestrally diverse populations: A transethnic genome-wide meta-analysis. PLoS Medicine, 2017, 14, e1002383.	3.9	341
107	The trans-ancestral genomic architecture of glycemic traits. Nature Genetics, 2021, 53, 840-860.	9.4	341
108	Genome-wide association study identifies a psoriasis susceptibility locus at TRAF3IP2. Nature Genetics, 2010, 42, 991-995.	9.4	331

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109	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. PLoS Genetics, 2015, 11, e1005378.	1.5	331
110	Strong Association of the Y402H Variant in Complement Factor H at 1q32 with Susceptibility to Age-Related Macular Degeneration. American Journal of Human Genetics, 2005, 77, 149-153.	2.6	327
111	Positional cloning of a novel gene influencing asthma from Chromosome 2q14. Nature Genetics, 2003, 35, 258-263.	9.4	326
112	Extent and Distribution of Linkage Disequilibrium in Three Genomic Regions. American Journal of Human Genetics, 2001, 68, 191-197.	2.6	325
113	CFH haplotypes without the Y402H coding variant show strong association with susceptibility to age-related macular degeneration. Nature Genetics, 2006, 38, 1049-1054.	9.4	318
114	Genome-wide association analysis identifies three psoriasis susceptibility loci. Nature Genetics, 2010, 42, 1000-1004.	9.4	313
115	A Comparison of Phasing Algorithms for Trios and Unrelated Individuals. American Journal of Human Genetics, 2006, 78, 437-450.	2.6	308
116	An efficient and scalable analysis framework for variant extraction and refinement from population-scale DNA sequence data. Genome Research, 2015, 25, 918-925.	2.4	308
117	Overexpression of the Cytokine BAFF and Autoimmunity Risk. New England Journal of Medicine, 2017, 376, 1615-1626.	13.9	301
118	Positional cloning of a quantitative trait locus on chromosome 13q14 that influences immunoglobulin E levels and asthma. Nature Genetics, 2003, 34, 181-186.	9.4	300
119	Combined Analysis of Genome-wide Association Studies for Crohn Disease and Psoriasis Identifies Seven Shared Susceptibility Loci. American Journal of Human Genetics, 2012, 90, 636-647.	2.6	290
120	Meta-analysis of Genome-wide Association Studies for Neuroticism, and the Polygenic Association With Major Depressive Disorder. JAMA Psychiatry, 2015, 72, 642.	6.0	289
121	New models of collaboration in genome-wide association studies: the Genetic Association Information Network. Nature Genetics, 2007, 39, 1045-1051.	9.4	288
122	Pedigree tests of transmission disequilibrium. European Journal of Human Genetics, 2000, 8, 545-551.	1.4	286
123	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	9.4	286
124	Genetic association study of QT interval highlights role for calcium signaling pathways in myocardial repolarization. Nature Genetics, 2014, 46, 826-836.	9.4	281
125	Powerful Regression-Based Quantitative-Trait Linkage Analysis of General Pedigrees. American Journal of Human Genetics, 2002, 71, 238-253.	2.6	276
126	Low-coverage sequencing: Implications for design of complex trait association studies. Genome Research, 2011, 21, 940-951.	2.4	273

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127	Punctuated bursts in human male demography inferred from 1,244 worldwide Y-chromosome sequences. Nature Genetics, 2016, 48, 593-599.	9.4	273
128	Systematic evaluation of coding variation identifies a candidate causal variant in TM6SF2 influencing total cholesterol and myocardial infarction risk. Nature Genetics, 2014, 46, 345-351.	9.4	268
129	Common Missense Variant in the Glucokinase Regulatory Protein Gene Is Associated With Increased Plasma Triglyceride and C-Reactive Protein but Lower Fasting Glucose Concentrations. Diabetes, 2008, 57, 3112-3121.	0.3	264
130	Unraveling a Multifactorial Late-Onset Disease: From Genetic Susceptibility to Disease Mechanisms for Age-Related Macular Degeneration. Annual Review of Genomics and Human Genetics, 2009, 10, 19-43.	2.5	254
131	Molecular Dissection of Psoriasis: Integrating Genetics and Biology. Journal of Investigative Dermatology, 2010, 130, 1213-1226.	0.3	253
132	Large scale meta-analysis characterizes genetic architecture for common psoriasis associated variants. Nature Communications, 2017, 8, 15382.	5.8	251
133	Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation. Nature Genetics, 2022, 54, 560-572.	9.4	250
134	The GLUT9 Gene Is Associated with Serum Uric Acid Levels in Sardinia and Chianti Cohorts. PLoS Genetics, 2007, 3, e194.	1.5	249
135	Exome sequencing of 20,791Âcases of type 2 diabetes and 24,440Âcontrols. Nature, 2019, 570, 71-76.	13.7	248
136	E2-2 Protein and Fuchs's Corneal Dystrophy. New England Journal of Medicine, 2010, 363, 1016-1024.	13.9	247
137	Exome array analysis identifies new loci and low-frequency variants influencing insulin processing and secretion. Nature Genetics, 2013, 45, 197-201.	9.4	247
138	Genetic loci influencing kidney function and chronic kidney disease. Nature Genetics, 2010, 42, 373-375.	9.4	246
139	Genome-wide Association Analysis of Psoriatic Arthritis and Cutaneous Psoriasis Reveals Differences in Their Genetic Architecture. American Journal of Human Genetics, 2015, 97, 816-836.	2.6	245
140	Handling Marker-Marker Linkage Disequilibrium: Pedigree Analysis with Clustered Markers. American Journal of Human Genetics, 2005, 77, 754-767.	2.6	239
141	Genotype-Imputation Accuracy across Worldwide Human Populations. American Journal of Human Genetics, 2009, 84, 235-250.	2.6	231
142	Population genetic differentiation of height and body mass index across Europe. Nature Genetics, 2015, 47, 1357-1362.	9.4	227
143	Genome-wide Association Study of Vitamin B6, Vitamin B12, Folate, and Homocysteine Blood Concentrations. American Journal of Human Genetics, 2009, 84, 477-482.	2.6	225
144	Meta-analysis of genome scans of age-related macular degeneration. Human Molecular Genetics, 2005, 14, 2257-2264.	1.4	224

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145	Genome-wide association study identifies variants in TMPRSS6 associated with hemoglobin levels. Nature Genetics, 2009, 41, 1170-1172.	9.4	217
146	A genome-wide association study on African-ancestry populations for asthma. Journal of Allergy and Clinical Immunology, 2010, 125, 336-346.e4.	1.5	213
147	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
148	Retinal transcriptome and eQTL analyses identify genes associated with age-related macular degeneration. Nature Genetics, 2019, 51, 606-610.	9.4	201
149	Whole-Exome Sequencing Identifies Rare and Low-Frequency Coding Variants Associated with LDL Cholesterol. American Journal of Human Genetics, 2014, 94, 233-245.	2.6	193
150	Genome sequencing elucidates Sardinian genetic architecture and augments association analyses for lipid and blood inflammatory markers. Nature Genetics, 2015, 47, 1272-1281.	9.4	193
151	Assessment of the Psoriatic Transcriptome in a Large Sample: Additional Regulated Genes and Comparisons with In Vitro Models. Journal of Investigative Dermatology, 2010, 130, 1829-1840.	0.3	192
152	Meta-analysis of genome-wide association studies from the CHARGE consortium identifies common variants associated with carotid intima media thickness and plaque. Nature Genetics, 2011, 43, 940-947.	9.4	191
153	Phenotypic Characterization of GeneticallyÂLowered Human Lipoprotein(a) Levels. Journal of the American College of Cardiology, 2016, 68, 2761-2772.	1.2	186
154	Genetic variation in the 22q11 locus and susceptibility to schizophrenia. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 16859-16864.	3.3	183
155	Fine Mapping Major Histocompatibility Complex Associations in Psoriasis and Its Clinical Subtypes. American Journal of Human Genetics, 2014, 95, 162-172.	2.6	182
156	Meta-analysis of gene-level tests for rare variant association. Nature Genetics, 2014, 46, 200-204.	9.4	178
157	Meta-analysis of Genome-Wide Association Studies for Extraversion: Findings from the Genetics of Personality Consortium. Behavior Genetics, 2016, 46, 170-182.	1.4	178
158	Toll-like receptor 4 variant D299G is associated with susceptibility to age-related macular degeneration. Human Molecular Genetics, 2005, 14, 1449-1455.	1.4	177
159	Whole-genome sequencing reveals host factors underlying critical COVID-19. Nature, 2022, 607, 97-103.	13.7	174
160	Gene Expression in Skin and Lymphoblastoid Cells: Refined Statistical Method Reveals Extensive Overlap in cis-eQTL Signals. American Journal of Human Genetics, 2010, 87, 779-789.	2.6	169
161	Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. Nature Communications, 2017, 8, 14977.	5.8	169
162	Genetic susceptibility to age-related macular degeneration: a paradigm for dissecting complex disease traits. Human Molecular Genetics, 2007, 16, R174-R182.	1.4	168

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163	Functional equivalence of genome sequencing analysis pipelines enables harmonized variant calling across human genetics projects. Nature Communications, 2018, 9, 4038.	5.8	166
164	Variants within the immunoregulatory CBLB gene are associated with multiple sclerosis. Nature Genetics, 2010, 42, 495-497.	9.4	164
165	Blood Pressure Loci Identified with a Gene-Centric Array. American Journal of Human Genetics, 2011, 89, 688-700.	2.6	159
166	Genotype Imputation from Large Reference Panels. Annual Review of Genomics and Human Genetics, 2018, 19, 73-96.	2.5	158
167	Genome-wide physical activity interactions in adiposity â€• A meta-analysis of 200,452 adults. PLoS Genetics, 2017, 13, e1006528.	1.5	158
168	Age-Related Macular Degeneration: A High-Resolution Genome Scan for Susceptibility Loci in a Population Enriched for Late-Stage Disease. American Journal of Human Genetics, 2004, 74, 482-494.	2.6	157
169	Enhanced meta-analysis and replication studies identify five new psoriasis susceptibility loci. Nature Communications, 2015, 6, 7001.	5.8	156
170	Assessing the contribution of rare variants to complex trait heritability from whole-genome sequence data. Nature Genetics, 2022, 54, 263-273.	9.4	156
171	Global Gene Expression Analysis Reveals Evidence for Decreased Lipid Biosynthesis and Increased Innate Immunity in Uninvolved Psoriatic Skin. Journal of Investigative Dermatology, 2009, 129, 2795-2804.	0.3	153
172	Complex genetic signatures in immune cells underlie autoimmunity and inform therapy. Nature Genetics, 2020, 52, 1036-1045.	9.4	153
173	LocusZoom.js: interactive and embeddable visualization of genetic association study results. Bioinformatics, 2021, 37, 3017-3018.	1.8	153
174	Identification of Novel Genetic Loci Associated with Thyroid Peroxidase Antibodies and Clinical Thyroid Disease. PLoS Genetics, 2014, 10, e1004123.	1.5	150
175	Association of Rare and Common Variation in the Lipoprotein Lipase Gene With Coronary Artery Disease. JAMA - Journal of the American Medical Association, 2017, 317, 937.	3.8	148
176	Association of Polygenic Risk Scores for Multiple Cancers in a Phenome-wide Study: Results from The Michigan Genomics Initiative. American Journal of Human Genetics, 2018, 102, 1048-1061.	2.6	147
177	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
178	Association Between Titin Loss-of-Function Variants and Early-Onset Atrial Fibrillation. JAMA - Journal of the American Medical Association, 2018, 320, 2354.	3.8	144
179	Genome-Wide Association Scan of Trait Depression. Biological Psychiatry, 2010, 68, 811-817.	0.7	143
180	A genome-wide association analysis of serum iron concentrations. Blood, 2010, 115, 94-96.	0.6	142

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181	Multiple Loci within the Major Histocompatibility Complex Confer Risk of Psoriasis. PLoS Genetics, 2009, 5, e1000606.	1.5	141
182	A Genome-Wide Association Scan on the Levels of Markers of Inflammation in Sardinians Reveals Associations That Underpin Its Complex Regulation. PLoS Genetics, 2012, 8, e1002480.	1.5	141
183	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals. Nature Communications, 2018, 9, 3391.	5.8	140
184	Amelioration of Sardinian Â0 thalassemia by genetic modifiers. Blood, 2009, 114, 3935-3937.	0.6	137
185	Improved Ancestry Estimation for both Genotyping and Sequencing Data using Projection Procrustes Analysis and Genotype Imputation. American Journal of Human Genetics, 2015, 96, 926-937.	2.6	137
186	Ancestry estimation and control of population stratification for sequence-based association studies. Nature Genetics, 2014, 46, 409-415.	9.4	136
187	A cross-platform analysis of 14,177 expression quantitative trait loci derived from lymphoblastoid cell lines. Genome Research, 2013, 23, 716-726.	2.4	135
188	Fine Mapping of Five Loci Associated with Low-Density Lipoprotein Cholesterol Detects Variants That Double the Explained Heritability. PLoS Genetics, 2011, 7, e1002198.	1.5	134
189	Elucidating the genetic architecture of familial schizophrenia using rare copy number variant and linkage scans. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 16746-16751.	3.3	133
190	Type 2 and interferon inflammation regulate SARS-CoV-2 entry factor expression in the airway epithelium. Nature Communications, 2020, 11, 5139.	5.8	131
191	Evidence of association ofAPOEwith age-related macular degeneration - a pooled analysis of 15 studies. Human Mutation, 2011, 32, 1407-1416.	1.1	130
192	Exome chip meta-analysis identifies novel loci and East Asianâ€“specific coding variants that contribute to lipid levels and coronary artery disease. Nature Genetics, 2017, 49, 1722-1730.	9.4	129
193	Exploring and visualizing large-scale genetic associations by using PheWeb. Nature Genetics, 2020, 52, 550-552.	9.4	129
194	In silico method for inferring genotypes in pedigrees. Nature Genetics, 2006, 38, 1002-1004.	9.4	127
195	The impact of genotyping error on family-based analysis of quantitative traits. European Journal of Human Genetics, 2001, 9, 130-134.	1.4	126
196	Common variants in the SLCO1B3 locus are associated with bilirubin levels and unconjugated hyperbilirubinemia. Human Molecular Genetics, 2009, 18, 2711-2718.	1.4	126
197	Phosphodiesterase 8B Gene Variants Are Associated with Serum TSH Levels and Thyroid Function. American Journal of Human Genetics, 2008, 82, 1270-1280.	2.6	124
198	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts. Nature Genetics, 2020, 52, 634-639.	9.4	124

#	Article	IF	Citations
199	Optimal designs for two-stage genome-wide association studies. Genetic Epidemiology, 2007, 31, 776-788.	0.6	123
200	Assessing Mitochondrial DNA Variation and Copy Number in Lymphocytes of ~2,000 Sardinians Using Tailored Sequencing Analysis Tools. PLoS Genetics, 2015, 11, e1005306.	1.5	123
201	Novel Blood Pressure Locus and Gene Discovery Using Genome-Wide Association Study and Expression Data Sets From Blood and the Kidney. Hypertension, 2017, 70, .	1.3	123
202	Whole genome sequencing in psychiatric disorders: the WGSPD consortium. Nature Neuroscience, 2017, 20, 1661-1668.	7.1	122
203	GENOME: a rapid coalescent-based whole genome simulator. Bioinformatics, 2007, 23, 1565-1567.	1.8	116
204	A Fast and Accurate Algorithm to Test for Binary Phenotypes and Its Application to PheWAS. American Journal of Human Genetics, 2017, 101, 37-49.	2.6	116
205	Joint Modeling of Linkage and Association: Identifying SNPs Responsible for a Linkage Signal. American Journal of Human Genetics, 2005, 76, 934-949.	2.6	114
206	Exome sequencing and complex disease: practical aspects of rare variant association studies. Human Molecular Genetics, 2012, 21, R1-R9.	1.4	114
207	52 Genetic Loci Influencing MyocardialÂMass. Journal of the American College of Cardiology, 2016, 68, 1435-1448.	1.2	113
208	RAREMETAL: fast and powerful meta-analysis for rare variants. Bioinformatics, 2014, 30, 2828-2829.	1.8	108
209	Genome-wide linkage analysis of a composite index of neuroticism and mood-related scales in extreme selected sibships. Human Molecular Genetics, 2004, 13, 2173-2182.	1.4	107
210	Efficient Study Designs for Test of Genetic Association Using Sibship Data and Unrelated Cases and Controls. American Journal of Human Genetics, 2006, 78, 778-792.	2.6	107
211	Functional Gene Group Analysis Reveals a Role of Synaptic Heterotrimeric G Proteins in Cognitive Ability. American Journal of Human Genetics, 2010, 86, 113-125.	2.6	106
212	Comprehensive Association Study of Type 2 Diabetes and Related Quantitative Traits With 222 Candidate Genes. Diabetes, 2008, 57, 3136-3144.	0.3	104
213	Genome-Wide Meta-Analysis of Psoriatic Arthritis Identifies Susceptibility Locus at REL. Journal of Investigative Dermatology, 2012, 132, 1133-1140.	0.3	99
214	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. Nature Communications, 2018, 9, 2252.	5.8	99
215	A comparison of approaches to account for uncertainty in analysis of imputed genotypes. Genetic Epidemiology, 2011, 35, 102-110.	0.6	98
216	Genome-wide association study of delay discounting in 23,217 adult research participants of European ancestry. Nature Neuroscience, 2018, 21, 16-18.	7.1	98

#	Article	IF	Citations
217	Genome-wide analysis provides genetic evidence that ACE2 influences COVID-19 risk and yields risk scores associated with severe disease. Nature Genetics, 2022, 54, 382-392.	9.4	97
218	Height-reducing variants and selection for short stature in Sardinia. Nature Genetics, 2015, 47, 1352-1356.	9.4	96
219	Genome-wide analysis of 53,400 people with irritable bowel syndrome highlights shared genetic pathways with mood and anxiety disorders. Nature Genetics, 2021, 53, 1543-1552.	9.4	96
220	The Power to Detect Linkage Disequilibrium with Quantitative Traits in Selected Samples. American Journal of Human Genetics, 2001, 68, 1463-1474.	2.6	95
221	Identification and Functional Characterization of G6PC2 Coding Variants Influencing Glycemic Traits Define an Effector Transcript at the G6PC2-ABCB11 Locus. PLoS Genetics, 2015, 11, e1004876.	1.5	95
222	Genetic signature to provide robust risk assessment of psoriatic arthritis development in psoriasis patients. Nature Communications, 2018, 9, 4178.	5.8	95
223	Rare variant genotype imputation with thousands of study-specific whole-genome sequences: implications for cost-effective study designs. European Journal of Human Genetics, 2015, 23, 975-983.	1.4	92
224	COL4A1 Is Associated With Arterial Stiffness by Genome-Wide Association Scan. Circulation: Cardiovascular Genetics, 2009, 2, 151-158.	5.1	91
225	Common Genetic Variation in the 3â€²- BCL11B Gene Desert Is Associated With Carotid-Femoral Pulse Wave Velocity and Excess Cardiovascular Disease Risk. Circulation: Cardiovascular Genetics, 2012, 5, 81-90.	5.1	90
226	Sequence features in regions of weak and strong linkage disequilibrium. Genome Research, 2005, 15, 1519-1534.	2.4	89
227	Mutations in a BTB-Kelch Protein, KLHL7, Cause Autosomal-Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 84, 792-800.	2.6	89
228	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. Nature Genetics, 2019, 51, 452-469.	9.4	89
229	Sex-dimorphic genetic effects and novel loci for fasting glucose and insulin variability. Nature Communications, 2021, 12, 24.	5.8	87
230	A functional IL-6 receptor (IL6R) variant is a risk factor for persistent atopic dermatitis. Journal of Allergy and Clinical Immunology, 2013, 132, 371-377.	1.5	86
231	Population Genomic Analysis of Ancient and Modern Genomes Yields New Insights into the Genetic Ancestry of the Tyrolean Iceman and the Genetic Structure of Europe. PLoS Genetics, 2014, 10, e1004353.	1.5	86
232	No Clinically Significant Association between CFH and ARMS2 Genotypes and Response to Nutritional Supplements. Ophthalmology, 2014, 121, 2173-2180.	2.5	86
233	Genome-wide Study of Atrial Fibrillation Identifies Seven Risk Loci and Highlights Biological Pathways and Regulatory Elements Involved in Cardiac Development. American Journal of Human Genetics, 2018, 102, 103-115.	2.6	86
234	Variations in Apolipoprotein E Frequency With Age in a Pooled Analysis of a Large Group of Older People. American Journal of Epidemiology, 2011, 173, 1357-1364.	1.6	85

#	Article	IF	Citations
235	Genomewide Scan in Families with Schizophrenia from the Founder Population of Afrikaners Reveals Evidence for Linkage and Uniparental Disomy on Chromosome 1. American Journal of Human Genetics, 2004, 74, 403-417.	2.6	84
236	Meta-analysis of up to 622,409 individuals identifies 40 novel smoking behaviour associated genetic loci. Molecular Psychiatry, 2020, 25, 2392-2409.	4.1	83
237	Platelet-Related Variants Identified by Exomechip Meta-analysis in 157,293 Individuals. American Journal of Human Genetics, 2016, 99, 40-55.	2.6	82
238	Toll-like Receptor Polymorphisms and Age-Related Macular Degeneration. , 2008, 49, 1652.		79
239	Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. International Journal of Epidemiology, 2012, 41, 250-262.	0.9	79
240	An alternative to the search for single polymorphisms: Toward molecular personality scales for the five-factor model.. Journal of Personality and Social Psychology, 2010, 99, 1014-1024.	2.6	76
241	Metabolic and cardiovascular traits: an abundance of recently identified common genetic variants. Human Molecular Genetics, 2008, 17, R102-R108.	1.4	75
242	Whole-genome sequence-based analysis of thyroid function. Nature Communications, 2015, 6, 5681.	5.8	75
243	An efficient comprehensive search algorithm for tagSNP selection using linkage disequilibrium criteria. Bioinformatics, 2006, 22, 220-225.	1.8	74
244	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. Nature Communications, 2016, 7, 13357.	5.8	74
245	Clonal hematopoiesis associated with epigenetic aging and clinical outcomes. Aging Cell, 2021, 20, e13366.	3.0	72
246	Pan-ancestry exome-wide association analyses of COVID-19 outcomes in 586,157 individuals. American Journal of Human Genetics, 2021, 108, 1350-1355.	2.6	72
247	A Likelihood-Based Framework for Variant Calling and De Novo Mutation Detection in Families. PLoS Genetics, 2012, 8, e1002944.	1.5	71
248	Increased Genetic Vulnerability to Smoking at CHRNA5 in Early-Onset Smokers. Archives of General Psychiatry, 2012, 69, 854.	13.8	71
249	Genomic history of the Sardinian population. Nature Genetics, 2018, 50, 1426-1434.	9.4	71
250	De novo mutations across 1,465 diverse genomes reveal mutational insights and reductions in the Amish founder population. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 2560-2569.	3.3	71
251	Nogo Receptor 1 (RTN4R) as a Candidate Gene for Schizophrenia: Analysis Using Human and Mouse Genetic Approaches. PLoS ONE, 2007, 2, e1234.	1.1	70
252	Exome Chip Meta-analysis Fine Maps Causal Variants and Elucidates the Genetic Architecture of Rare Coding Variants in Smoking and AlcoholÂUse. Biological Psychiatry, 2019, 85, 946-955.	0.7	69

#	Article	IF	Citations
253	A high-resolution HLA reference panel capturing global population diversity enables multi-ancestry fine-mapping in HIV host response. Nature Genetics, 2021, 53, 1504-1516.	9.4	69
254	Prosaposin is a regulator of progranulin levels and oligomerization. Nature Communications, 2016, 7, 11992.	5.8	68
255	Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 individuals in the UK Biobank. Nature Genetics, 2022, 54, 240-250.	9.4	68
256	Genome-wide analysis of disease progression in age-related macular degeneration. Human Molecular Genetics, 2018, 27, 929-940.	1.4	67
257	Association between quantitative traits underlying asthma and the HLA-DRB1 locus in a family-based population sample. European Journal of Human Genetics, 2001, 9, 341-346.	1.4	66
258	Genome-wide association analyses based on whole-genome sequencing in Sardinia provide insights into regulation of hemoglobin levels. Nature Genetics, 2015, 47, 1264-1271.	9.4	66
259	Data for Genetic Analysis Workshop 18: human whole genome sequence, blood pressure, and simulated phenotypes in extended pedigrees. BMC Proceedings, 2014, 8, S2.	1.8	65
260	A Scalable Bayesian Method for Integrating Functional Information in Genome-wide Association Studies. American Journal of Human Genetics, 2017, 101, 404-416.	2.6	63
261	Increasing the Power and Efficiency of Disease-Marker Case-Control Association Studies through Use of Allele-Sharing Information. American Journal of Human Genetics, 2004, 74, 432-443.	2.6	62
262	Mitogenome Diversity in Sardinians: A Genetic Window onto an Island's Past. Molecular Biology and Evolution, 2017, 34, 1230-1239.	3.5	61
263	Genetic Testing in Persons with Age-Related Macular Degeneration and the Use of the AREDS Supplements: To Test or Not to Test?. Ophthalmology, 2015, 122, 212-215.	2.5	58
264	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
265	Sex-specific and pleiotropic effects underlying kidney function identified from GWAS meta-analysis. Nature Communications, 2019, 10, 1847.	5.8	55
266	Ancestry-agnostic estimation of DNA sample contamination from sequence reads. Genome Research, 2020, 30, 185-194.	2.4	51
267	Inflammation in the pathogenesis of age-related macular degeneration. British Journal of Ophthalmology, 2008, 92, 448-450.	2.1	50
268	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
269	A 32 kb Critical Region Excluding Y402H in CFH Mediates Risk for Age-Related Macular Degeneration. PLoS ONE, 2011, 6, e25598.	1.1	46
270	Genotype calling and haplotyping in parent-offspring trios. Genome Research, 2013, 23, 142-151.	2.4	46

#	Article	IF	Citations
271	Bivariate Analysis of Age-Related Macular Degeneration Progression Using Genetic Risk Scores. Genetics, 2017, 206, 119-133.	1.2	46
272	Linkage Disequilibrium: Ancient History Drives the New Genetics. Human Heredity, 2005, 59, 118-124.	0.4	45
273	Genetic architectures of proximal and distal colorectal cancer are partly distinct. Gut, 2021, 70, 1325-1334.	6.1	44
274	Characterization of ADME gene variation in 21 populations by exome sequencing. Pharmacogenetics and Genomics, 2017, 27, 89-100.	0.7	43
275	The Molecular Genetic Architecture of Self-Employment. PLoS ONE, 2013, 8, e60542.	1.1	41
276	Rare Nonsynonymous Exonic Variants in Addiction and Behavioral Disinhibition. Biological Psychiatry, 2014, 75, 783-789.	0.7	41
277	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling. Human Molecular Genetics, 2017, 26, 4301-4313.	1.4	41
278	Association Analysis in a Variance Components Framework. Genetic Epidemiology, 2001, 21, S341-6.	0.6	40
279	Genotypeâ€based matching to correct for population stratification in largeâ€scale caseâ€control genetic association studies. Genetic Epidemiology, 2009, 33, 508-517.	0.6	40
280	Fine Mapping on Chromosome 13q32â€“34 and Brain Expression Analysis Implicates MYO16 in Schizophrenia. Neuropsychopharmacology, 2014, 39, 934-943.	2.8	40
281	Correcting for Sample Contamination in Genotype Calling of DNA Sequence Data. American Journal of Human Genetics, 2015, 97, 284-290.	2.6	39
282	Loss-of-function genomic variants highlight potential therapeutic targets for cardiovascular disease. Nature Communications, 2020, 11, 6417.	5.8	39
283	Population- and individual-specific regulatory variation in Sardinia. Nature Genetics, 2017, 49, 700-707.	9.4	38
284	Some properties of a variance components model for fine-mapping quantitative trait loci. , 2000, 30, 235-243.		36
285	Variants of the serotonin transporter gene and NEOâ€PIâ€R Neuroticism: No association in the BLSA and SardiNIA samples. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 1070-1077.	1.1	36
286	Prevalence of CKD and Its Relationship to eGFR-Related Genetic Loci and Clinical Risk Factors in the SardiNIA Study Cohort. Journal of the American Society of Nephrology: JASN, 2014, 25, 1533-1544.	3.0	36
287	Imputation-Aware Tag SNP Selection To Improve Power for Large-Scale, Multi-ethnic Association Studies. G3: Genes, Genomes, Genetics, 2018, 8, 3255-3267.	0.8	36
288	Identifying Novel Susceptibility Genes for Colorectal Cancer Risk From a Transcriptome-Wide Association Study of 125,478 Subjects. Gastroenterology, 2021, 160, 1164-1178.e6.	0.6	36

#	Article	IF	Citations
289	Mendelian randomization supports bidirectional causality between telomere length and clonal hematopoiesis of indeterminate potential. Science Advances, 2022, 8, eabl6579.	4.7	36
290	Genetic association study of age-related macular degeneration in the Spanish population. Acta Ophthalmologica, 2011, 89, e12-e22.	0.6	33
291	Genetic variants inCETPincrease risk of intracerebral hemorrhage. Annals of Neurology, 2016, 80, 730-740.	2.8	33
292	Whole genome sequence analysis of pulmonary function and COPD in 19,996 multi-ethnic participants. Nature Communications, 2020, 11, 5182.	5.8	32
293	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. Scientific Data, 2017, 4, 170179.	2.4	31
294	LASER server: ancestry tracing with genotypes or sequence reads. Bioinformatics, 2017, 33, 2056-2058.	1.8	30
295	A Mixed-Effects Model for Powerful Association Tests in Integrative Functional Genomics. American Journal of Human Genetics, 2018, 102, 904-919.	2.6	30
296	A Novel Recurrent COL5A1 Genetic Variant Is Associated With a Dysplasia-Associated Arterial Disease Exhibiting Dissections and Fibromuscular Dysplasia. Arteriosclerosis, Thrombosis, and Vascular Biology, 2020, 40, 2686-2699.	1.1	30
297	Psoriasis Bench to Bedside. Archives of Dermatology, 2009, 145, 462-4.	1.7	29
298	Genome sequencing unveils a regulatory landscape of platelet reactivity. Nature Communications, 2021, 12, 3626.	5.8	29
299	Genetic determinants of telomere length from 109,122 ancestrally diverse whole-genome sequences in TOPMed. Cell Genomics, 2022, 2, 100084.	3.0	29
300	Estimating the power of variance component linkage analysis in large pedigrees. Genetic Epidemiology, 2006, 30, 471-484.	0.6	28
301	Exploring various polygenic risk scores for skin cancer in the phenomes of the Michigan genomics initiative and the UK Biobank with a visual catalog: PRSWeb. PLoS Genetics, 2019, 15, e1008202.	1.5	28
302	Quantitative Trait Linkage Analysis Using Gaussian Copulas. Genetics, 2006, 173, 2317-2327.	1.2	27
303	Improving power of association tests using multiple sets of imputed genotypes from distributed reference panels. Genetic Epidemiology, 2017, 41, 744-755.	0.6	27
304	Age-of-onset information helps identify 76 genetic variants associated with allergic disease. PLoS Genetics, 2020, 16, e1008725.	1.5	27
305	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. Human Molecular Genetics, 2020, 29, 2022-2034.	1.4	26
306	Association of mitochondrial DNA copy number with cardiometabolic diseases. Cell Genomics, 2021, 1, 100006.	3.0	26

#	Article	IF	Citations
307	Clonal hematopoiesis in sickle cell disease. Journal of Clinical Investigation, 2022, 132, .	3.9	26
308	In search of rare variants: Preliminary results from whole genome sequencing of 1,325 individuals with psychophysiological endophenotypes. Psychophysiology, 2014, 51, 1309-1320.	1.2	25
309	Fine mapping of eight psoriasis susceptibility loci. European Journal of Human Genetics, 2015, 23, 844-853.	1.4	25
310	Protein Tyrosine Phosphatase Gene PTPN22 Polymorphism in Psoriasis: Lack of Evidence for Association. Journal of Investigative Dermatology, 2005, 125, 395-396.	0.3	24
311	Refining genome-wide linkage intervals using a meta-analysis of genome-wide association studies identifies loci influencing personality dimensions. European Journal of Human Genetics, 2013, 21, 876-882.	1.4	24
312	MEPE loss-of-function variant associates with decreased bone mineral density and increased fracture risk. Nature Communications, 2020, 11, 4093.	5.8	24
313	Nasal airway transcriptome-wide association study of asthma reveals genetically driven mucus pathobiology. Nature Communications, 2022, 13, 1632.	5.8	24
314	The Effect of Genotype and Pedigree Error on Linkage Analysis: Analysis of Three Asthma Genome Scans. Genetic Epidemiology, 2001, 21, S117-22.	0.6	21
315	Genetic associations of nonsynonymous exonic variants with psychophysiological endophenotypes. Psychophysiology, 2014, 51, 1300-1308.	1.2	21
316	Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal regulatory mechanisms. Human Molecular Genetics, 2016, 25, 2070-2081.	1.4	21
317	Narrow-sense heritability estimation of complex traits using identity-by-descent information. Heredity, 2018, 121, 616-630.	1.2	20
318	Using sex-averaged genetic maps in multipoint linkage analysis when identity-by-descent status is incompletely known. Genetic Epidemiology, 2006, 30, 384-396.	0.6	19
319	Optimal sequencing strategies for identifying disease-associated singletons. PLoS Genetics, 2017, 13, e1006811.	1.5	19
320	Proper conditional analysis in the presence of missing data: Application to large scale meta-analysis of tobacco use phenotypes. PLoS Genetics, 2018, 14, e1007452.	1.5	18
321	Omics-squared: human genomic, transcriptomic and phenotypic data for genetic analysis workshop 19. BMC Proceedings, 2016, 10, 71-77.	1.8	17
322	Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. Nature Communications, 2021, 12, 2182.	5.8	17
323	The Benefits of Using Genetic Information to Design Prevention Trials. American Journal of Human Genetics, 2013, 92, 547-557.	2.6	16
324	No large-effect low-frequency coding variation found for myocardial infarction. Human Molecular Genetics, 2014, 23, 4721-4728.	1.4	16

#	Article	IF	Citations
325	Genes for Good: Engaging the Public in Genetics Research via Social Media. American Journal of Human Genetics, 2019, 105, 65-77.	2.6	16
326	Mapping the 17q12â€“21.1 Locus for Variants Associated with Early-Onset Asthma in African Americans. American Journal of Respiratory and Critical Care Medicine, 2021, 203, 424-436.	2.5	16
327	Whole-genome association analyses of sleep-disordered breathing phenotypes in the NHLBI TOPMed program. Genome Medicine, 2021, 13, 136.	3.6	16
328	Asthma and its relationship to mitochondrial copy number: Results from the Asthma Translational Genomics Collaborative (ATGC) of the Trans-Omics for Precision Medicine (TOPMed) program. PLoS ONE, 2020, 15, e0242364.	1.1	16
329	emeraLD: rapid linkage disequilibrium estimation with massive datasets. Bioinformatics, 2019, 35, 164-166.	1.8	15
330	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. Molecular Psychiatry, 2021, 26, 5239-5250.	4.1	15
331	GWAS of stool frequency provides insights into gastrointestinal motility and irritable bowel syndrome. Cell Genomics, 2021, 1, 100069.	3.0	15
332	FIVEx: an interactive eQTL browser across public datasets. Bioinformatics, 2022, 38, 559-561.	1.8	14
333	Whole-genome sequencing in diverse subjects identifies genetic correlates of leukocyte traits: The NHLBI TOPMed program. American Journal of Human Genetics, 2021, 108, 1836-1851.	2.6	14
334	LabWAS: Novel findings and study design recommendations from a meta-analysis of clinical labs in two independent biobanks. PLoS Genetics, 2020, 16, e1009077.	1.5	14
335	AbCD: arbitrary coverage design for sequencing-based genetic studies. Bioinformatics, 2013, 29, 799-801.	1.8	13
336	PRF1 mutation alters immune system activation, inflammation, and risk of autoimmunity. Multiple Sclerosis Journal, 2021, 27, 1332-1340.	1.4	13
337	Genome-wide association study of susceptibility loci for breast cancer in Sardinian population. BMC Cancer, 2015, 15, 383.	1.1	12
338	Association of Rare Predicted Loss-of-Function Variants in Cellular Pathways with Sub-Phenotypes in Age-Related Macular Degeneration. Ophthalmology, 2018, 125, 398-406.	2.5	12
339	Population-scale analysis of common and rare genetic variation associated with hearing loss in adults. Communications Biology, 2022, 5, .	2.0	12
340	Insights From a Large-Scale Whole-Genome Sequencing Study of Systolic Blood Pressure, Diastolic Blood Pressure, and Hypertension. Hypertension, 2022, 79, 1656-1667.	1.3	12
341	SHAVE: shrinkage estimator measured for multiple visits increases power in GWAS of quantitative traits. European Journal of Human Genetics, 2013, 21, 673-679.	1.4	11
342	Targeted exonic sequencing of GWAS loci in the high extremes of the plasma lipids distribution. Atherosclerosis, 2016, 250, 63-68.	0.4	11

#	Article	IF	Citations
343	Estimation of DNA contamination and its sources in genotyped samples. Genetic Epidemiology, 2019, 43, 980-995.	0.6	11
344	Lung Function in African American Children with Asthma Is Associated with Novel Regulatory Variants of the KIT LigandKITLG/SCFand Gene-By-Air-Pollution Interaction. Genetics, 2020, 215, 869-886.	1.2	11
345	Whole-Genome Sequencing Identifies Novel Functional Loci Associated with Lung Function in Puerto Rican Youth. American Journal of Respiratory and Critical Care Medicine, 2020, 202, 962-972.	2.5	11
346	Integrating comprehensive functional annotations to boost power and accuracy in gene-based association analysis. PLoS Genetics, 2020, 16, e1009060.	1.5	11
347	Genome-wide meta-analysis of iron status biomarkers and the effect of iron on all-cause mortality in HUNT. Communications Biology, 2022, 5, .	2.0	11
348	Graphical algorithm for integration of genetic and biological data: proof of principle using psoriasis as a model. Bioinformatics, 2015, 31, 1243-1249.	1.8	10
349	Metaâ€MultiSKAT: Multiple phenotype metaâ€analysis for regionâ€based association test. Genetic Epidemiology, 2019, 43, 800-814.	0.6	9
350	Relative impact of indels versus SNPs on complex disease. Genetic Epidemiology, 2019, 43, 112-117.	0.6	9
351	Whole genome sequence analysis of platelet traits in the NHLBI Trans-Omics for Precision Medicine (TOPMed) initiative. Human Molecular Genetics, 2022, 31, 347-361.	1.4	9
352	GWAS GUI: graphical browser for the results of whole-genome association studies with high-dimensional phenotypes. Bioinformatics, 2009, 25, 284-285.	1.8	8
353	A Single SNP Surrogate for Genotyping HLA-C*06:02 in Diverse Populations. Journal of Investigative Dermatology, 2015, 135, 1177-1180.	0.3	8
354	Fine-Mapping of Common Genetic Variants Associated with Colorectal Tumor Risk Identified Potential Functional Variants. PLoS ONE, 2016, 11, e0157521.	1.1	8
355	Transethnic analysis of psoriasis susceptibility in South Asians and Europeans enhances fine mapping in the MHC and genome wide. Human Genetics and Genomics Advances, 2022, 3, 100069.	1.0	8
356	An evaluation of the replicate pool method: quick estimation of genome-wide linkage peakp-values. Genetic Epidemiology, 2006, 30, 320-332.	0.6	7
357	Impact of natural selection on global patterns of genetic variation and association with clinical phenotypes at genes involved in SARS-CoV-2 infection. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2123000119.	3.3	7
358	Regression-Based Sib Pair Linkage Analysis for Binary Traits. Human Heredity, 2003, 55, 125-131.	0.4	6
359	Improved score statistics for metaâ€analysis in singleâ€variant and geneâ€level association studies. Genetic Epidemiology, 2018, 42, 333-343.	0.6	6
360	Robust, flexible, and scalable tests for Hardyâ€“Weinberg equilibrium across diverse ancestries. Genetics, 2021, 218, .	1.2	6

#	Article	IF	Citations
361	Presence and transmission of mitochondrial heteroplasmic mutations in human populations of European and African ancestry. Mitochondrion, 2021, 60, 33-42.	1.6	6
362	Enhanced genetic maps from family-based disease studies: population-specific comparisons. BMC Medical Genetics, 2011, 12, 15.	2.1	5
363	Rare variant associations with waist-to-hip ratio in European-American and African-American women from the NHLBI-Exome Sequencing Project. European Journal of Human Genetics, 2016, 24, 1181-1187.	1.4	5
364	Identification of CFTR variants in Latino patients with cystic fibrosis from the Dominican Republic and Puerto Rico. Pediatric Pulmonology, 2020, 55, 533-540.	1.0	5
365	Variance Components Linkage Analysis with Repeated Measurements. Human Heredity, 2009, 67, 237-247.	0.4	4
366	Association Analysis and Meta-Analysis of Multi-Allelic Variants for Large-Scale Sequence Data. Genes, 2020, 11, 586.	1.0	3
367	Sparse allele vectors and the savvy software suite. Bioinformatics, 2021, 37, 4248-4250.	1.8	3
368	Response to Graffelman: Tests of Hardy-Weinberg Equilibrium. American Journal of Human Genetics, 2010, 86, 818-819.	2.6	2
369	A Sardinian founder mutation in glycoprotein Ib platelet subunit beta (GP1BB) that impacts thrombocytopenia. British Journal of Haematology, 2020, 191, e124-e128.	1.2	2
370	Regression models for association studies of quantitative trait loci in humans. GeneScreen, 2000, 1, 55-57.	0.7	1
371	Genome-wide Association Study of Vitamin B6, Vitamin B12, Folate, and Homocysteine Blood Concentrations. American Journal of Human Genetics, 2009, 84, 712.	2.6	1
372	Author reply. Ophthalmology, 2015, 122, e46-e47.	2.5	1
373	Independent test assessment using the extreme value distribution theory. BMC Proceedings, 2016, 10, 245-249.	1.8	1
374	The GLUT9 Gene is Associated with Serum Uric Acid Levels in Sardinia and Chianti Cohorts. PLoS Genetics, 2005, preprint, e194.	1.5	1
375	UGT1A1 genetic variants are associated with increases in bilirubin levels in rheumatoid arthritis patients treated with sarilumab. Pharmacogenomics Journal, 2022, 22, 160-165.	0.9	1
376	Reply. Ophthalmology, 2015, 122, e63.	2.5	0
377	Reply. Ophthalmology, 2015, 122, e58-e59.	2.5	0
378	2014 Curt Stern Award: Adventures in Human Genetics1. American Journal of Human Genetics, 2015, 96, 363-366.	2.6	0

#	Article	IF	Citations
379	Reply. Ophthalmology, 2015, 122, e61-e62.	2.5	0
380	Genetic Modifiers of Homozygous Beta Zero Thalassemia.. Blood, 2008, 112, 1874-1874.	0.6	0