

Jonathan K Pritchard

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

134
papers

69,444
citations

82
h-index

155
g-index

155
ext. papers

81,506
ext. citations

20.1
avg, IF

8.11
L-index

#	Paper	IF	Citations
134	Inference of population structure using multilocus genotype data. <i>Genetics</i> , 2000 , 155, 945-59	4	22315
133	Inference of population structure using multilocus genotype data: linked loci and correlated allele frequencies. <i>Genetics</i> , 2003 , 164, 1567-87	4	5901
132	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans. <i>Science</i> , 2015 , 348, 648-60	33.3	3242
131	Inferring weak population structure with the assistance of sample group information. <i>Molecular Ecology Resources</i> , 2009 , 9, 1322-32	8.4	2406
130	Inference of population structure using multilocus genotype data: dominant markers and null alleles. <i>Molecular Ecology Notes</i> , 2007 , 7, 574-578		2333
129	Genetic structure of human populations. <i>Science</i> , 2002 , 298, 2381-5	33.3	1995
128	A map of recent positive selection in the human genome. <i>PLoS Biology</i> , 2006 , 4, e72	9.7	1811
127	Association mapping in structured populations. <i>American Journal of Human Genetics</i> , 2000 , 67, 170-81	11	1472
126	An Expanded View of Complex Traits: From Polygenic to Omnigenic. <i>Cell</i> , 2017 , 169, 1177-1186	56.2	1413
125	Are rare variants responsible for susceptibility to complex diseases?. <i>American Journal of Human Genetics</i> , 2001 , 69, 124-37	11	1171
124	Convergent adaptation of human lactase persistence in Africa and Europe. <i>Nature Genetics</i> , 2007 , 39, 31-40	36.3	1073
123	Understanding mechanisms underlying human gene expression variation with RNA sequencing. <i>Nature</i> , 2010 , 464, 768-72	50.4	993
122	Use of unlinked genetic markers to detect population stratification in association studies. <i>American Journal of Human Genetics</i> , 1999 , 65, 220-8	11	993
121	Linkage disequilibrium in humans: models and data. <i>American Journal of Human Genetics</i> , 2001 , 69, 1-14	11	920
120	A systematic survey of loss-of-function variants in human protein-coding genes. <i>Science</i> , 2012 , 335, 823-833	33.3	880
119	fastSTRUCTURE: variational inference of population structure in large SNP data sets. <i>Genetics</i> , 2014 , 197, 573-89	4	839
118	Traces of human migrations in <i>Helicobacter pylori</i> populations. <i>Science</i> , 2003 , 299, 1582-5	33.3	764

117	The genetics of human adaptation: hard sweeps, soft sweeps, and polygenic adaptation. <i>Current Biology</i> , 2010 , 20, R208-15	6.3	666
116	DNA methylation patterns associate with genetic and gene expression variation in HapMap cell lines. <i>Genome Biology</i> , 2011 , 12, R10	18.3	651
115	Signals of recent positive selection in a worldwide sample of human populations. <i>Genome Research</i> , 2009 , 19, 826-37	9.7	557
114	Lineage-specific and single-cell chromatin accessibility charts human hematopoiesis and leukemia evolution. <i>Nature Genetics</i> , 2016 , 48, 1193-203	36.3	555
113	A high-resolution survey of deletion polymorphism in the human genome. <i>Nature Genetics</i> , 2006 , 38, 75-81	36.3	539
112	The allelic architecture of human disease genes: common disease-common variant...or not?. <i>Human Molecular Genetics</i> , 2002 , 11, 2417-23	5.6	515
111	Using environmental correlations to identify loci underlying local adaptation. <i>Genetics</i> , 2010 , 185, 1411-23	23	497
110	Informativeness of genetic markers for inference of ancestry. <i>American Journal of Human Genetics</i> , 2003 , 73, 1402-22	11	495
109	DNase I sensitivity QTLs are a major determinant of human expression variation. <i>Nature</i> , 2012 , 482, 390-4	40.4	479
108	High-resolution mapping of expression-QTLs yields insight into human gene regulation. <i>PLoS Genetics</i> , 2008 , 4, e1000214	6	456
107	Haplotype blocks and linkage disequilibrium in the human genome. <i>Nature Reviews Genetics</i> , 2003 , 4, 587-97	30.1	430
106	Sequencing and analysis of Neanderthal genomic DNA. <i>Science</i> , 2006 , 314, 1113-8	33.3	415
105	Clines, clusters, and the effect of study design on the inference of human population structure. <i>PLoS Genetics</i> , 2005 , 1, e70	6	407
104	Accurate inference of transcription factor binding from DNA sequence and chromatin accessibility data. <i>Genome Research</i> , 2011 , 21, 447-55	9.7	406
103	A worldwide survey of haplotype variation and linkage disequilibrium in the human genome. <i>Nature Genetics</i> , 2006 , 38, 1251-60	36.3	406
102	Effect of read-mapping biases on detecting allele-specific expression from RNA-sequencing data. <i>Bioinformatics</i> , 2009 , 25, 3207-12	7.2	390
101	Revealing the architecture of gene regulation: the promise of eQTL studies. <i>Trends in Genetics</i> , 2008 , 24, 408-15	8.5	366
100	RNA splicing is a primary link between genetic variation and disease. <i>Science</i> , 2016 , 352, 600-4	33.3	357

99	Tracing the peopling of the world through genomics. <i>Nature</i> , 2017 , 541, 302-310	50.4	338
98	DNA sequence-dependent compartmentalization and silencing of chromatin at the nuclear lamina. <i>Cell</i> , 2012 , 149, 1474-87	56.2	337
97	Identification of genetic variants that affect histone modifications in human cells. <i>Science</i> , 2013 , 342, 747-9	33.3	331
96	Case-control studies of association in structured or admixed populations. <i>Theoretical Population Biology</i> , 2001 , 60, 227-37	1.2	325
95	Adaptation - not by sweeps alone. <i>Nature Reviews Genetics</i> , 2010 , 11, 665-7	30.1	321
94	The role of geography in human adaptation. <i>PLoS Genetics</i> , 2009 , 5, e1000500	6	304
93	Clonal origin and evolution of a transmissible cancer. <i>Cell</i> , 2006 , 126, 477-87	56.2	299
92	Overcoming the winner's curse: estimating penetrance parameters from case-control data. <i>American Journal of Human Genetics</i> , 2007 , 80, 605-15	11	289
91	High-resolution mapping of crossovers reveals extensive variation in fine-scale recombination patterns among humans. <i>Science</i> , 2008 , 319, 1395-8	33.3	287
90	Genomic variation. Impact of regulatory variation from RNA to protein. <i>Science</i> , 2015 , 347, 664-7	33.3	276
89	WASP: allele-specific software for robust molecular quantitative trait locus discovery. <i>Nature Methods</i> , 2015 , 12, 1061-3	21.6	253
88	Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions. <i>Cell</i> , 2015 , 162, 1051-65	56.2	240
87	Annotation-free quantification of RNA splicing using LeafCutter. <i>Nature Genetics</i> , 2018 , 50, 151-158	36.3	234
86	Detection of human adaptation during the past 2000 years. <i>Science</i> , 2016 , 354, 760-764	33.3	224
85	The deleterious mutation load is insensitive to recent population history. <i>Nature Genetics</i> , 2014 , 46, 220-4	36.3	214
84	Adaptations to climate-mediated selective pressures in humans. <i>PLoS Genetics</i> , 2011 , 7, e1001375	6	207
83	Batch effects and the effective design of single-cell gene expression studies. <i>Scientific Reports</i> , 2017 , 7, 39921	4.9	205
82	Adaptations to climate in candidate genes for common metabolic disorders. <i>PLoS Genetics</i> , 2008 , 4, e32	6	204

81	Noisy splicing drives mRNA isoform diversity in human cells. <i>PLoS Genetics</i> , 2010 , 6, e1001236	6	203
80	Controls of nucleosome positioning in the human genome. <i>PLoS Genetics</i> , 2012 , 8, e1003036	6	196
79	Genomics: ENCODE explained. <i>Nature</i> , 2012 , 489, 52-5	50.4	191
78	Methylation QTLs are associated with coordinated changes in transcription factor binding, histone modifications, and gene expression levels. <i>PLoS Genetics</i> , 2014 , 10, e1004663	6	187
77	Abundant contribution of short tandem repeats to gene expression variation in humans. <i>Nature Genetics</i> , 2016 , 48, 22-9	36.3	184
76	Trans Effects on Gene Expression Can Drive Omnigenic Inheritance. <i>Cell</i> , 2019 , 177, 1022-1034.e6	56.2	183
75	Reduced signal for polygenic adaptation of height in UK Biobank. <i>ELife</i> , 2019 , 8,	8.9	181
74	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , 2015 , 348, 666-9	33.3	170
73	Confounding from cryptic relatedness in case-control association studies. <i>PLoS Genetics</i> , 2005 , 1, e32	6	163
72	A genome-wide study of DNA methylation patterns and gene expression levels in multiple human and chimpanzee tissues. <i>PLoS Genetics</i> , 2011 , 7, e1001316	6	159
71	Primate transcript and protein expression levels evolve under compensatory selection pressures. <i>Science</i> , 2013 , 342, 1100-4	33.3	156
70	Completing the map of human genetic variation. <i>Nature</i> , 2007 , 447, 161-5	50.4	153
69	Dissecting the regulatory architecture of gene expression QTLs. <i>Genome Biology</i> , 2012 , 13, R7	18.3	151
68	The genetic architecture of adaptations to high altitude in Ethiopia. <i>PLoS Genetics</i> , 2012 , 8, e1003110	6	137
67	The functional consequences of variation in transcription factor binding. <i>PLoS Genetics</i> , 2014 , 10, e1004226	13.5	135
66	Admixture facilitates genetic adaptations to high altitude in Tibet. <i>Nature Communications</i> , 2014 , 5, 3281	17.4	133
65	Statistical tests for admixture mapping with case-control and cases-only data. <i>American Journal of Human Genetics</i> , 2004 , 75, 771-89	11	132
64	Assessing the performance of the haplotype block model of linkage disequilibrium. <i>American Journal of Human Genetics</i> , 2003 , 73, 502-15	11	124

63	Variable prediction accuracy of polygenic scores within an ancestry group. <i>ELife</i> , 2020 , 9,	8.9	112
62	Characterizing natural variation using next-generation sequencing technologies. <i>Trends in Genetics</i> , 2009 , 25, 463-71	8.5	108
61	Coalescent-based association mapping and fine mapping of complex trait loci. <i>Genetics</i> , 2005 , 169, 1071-92	9.2	99
60	Comparative RNA sequencing reveals substantial genetic variation in endangered primates. <i>Genome Research</i> , 2012 , 22, 602-10	9.7	97
59	The genetic and mechanistic basis for variation in gene regulation. <i>PLoS Genetics</i> , 2015 , 11, e1004857	6	96
58	Evidence for extensive transmission distortion in the human genome. <i>American Journal of Human Genetics</i> , 2004 , 74, 62-72	11	94
57	Coregulation of tandem duplicate genes slows evolution of subfunctionalization in mammals. <i>Science</i> , 2016 , 352, 1009-13	33.3	90
56	Functional Genetic Variants Revealed by Massively Parallel Precise Genome Editing. <i>Cell</i> , 2018 , 175, 544-562.e16	56.7	89
55	Landscape of stimulation-responsive chromatin across diverse human immune cells. <i>Nature Genetics</i> , 2019 , 51, 1494-1505	36.3	86
54	Rapid evolution of the human mutation spectrum. <i>ELife</i> , 2017 , 6,	8.9	82
53	Thousands of novel translated open reading frames in humans inferred by ribosome footprint profiling. <i>ELife</i> , 2016 , 5,	8.9	82
52	The contribution of RNA decay quantitative trait loci to inter-individual variation in steady-state gene expression levels. <i>PLoS Genetics</i> , 2012 , 8, e1003000	6	80
51	Genetic Variation, Not Cell Type of Origin, Underlies the Majority of Identifiable Regulatory Differences in iPSCs. <i>PLoS Genetics</i> , 2016 , 12, e1005793	6	79
50	Genetic variation in MHC proteins is associated with T cell receptor expression biases. <i>Nature Genetics</i> , 2016 , 48, 995-1002	36.3	79
49	Genetics of 35 blood and urine biomarkers in the UK Biobank. <i>Nature Genetics</i> , 2021 , 53, 185-194	36.3	78
48	Large-Scale Clonal Analysis Resolves Aging of the Mouse Hematopoietic Stem Cell Compartment. <i>Cell Stem Cell</i> , 2018 , 22, 600-607.e4	18	77
47	Chromatin accessibility dynamics in a model of human forebrain development. <i>Science</i> , 2020 , 367,	33.3	76
46	Gene expression levels are a target of recent natural selection in the human genome. <i>Molecular Biology and Evolution</i> , 2009 , 26, 649-58	8.3	73

45	Genome-wide association study of behavioral, physiological and gene expression traits in outbred CFW mice. <i>Nature Genetics</i> , 2016 , 48, 919-26	36.3	72
44	Impact of regulatory variation across human iPSCs and differentiated cells. <i>Genome Research</i> , 2018 , 28, 122-131	9.7	70
43	Inferring Relevant Cell Types for Complex Traits by Using Single-Cell Gene Expression. <i>American Journal of Human Genetics</i> , 2017 , 101, 686-699	11	61
42	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. <i>Nature Genetics</i> , 2021 , 53, 1300-1310	36.3	60
41	Ancient Rome: A genetic crossroads of Europe and the Mediterranean. <i>Science</i> , 2019 , 366, 708-714	33.3	59
40	Determining the genetic basis of anthracycline-cardiotoxicity by molecular response QTL mapping in induced cardiomyocytes. <i>ELife</i> , 2018 , 7,	8.9	54
39	Interpreting polygenic scores, polygenic adaptation, and human phenotypic differences. <i>Evolution, Medicine and Public Health</i> , 2019 , 2019, 26-34	3	51
38	Evolutionary Persistence of DNA Methylation for Millions of Years after Ancient Loss of a De Novo Methyltransferase. <i>Cell</i> , 2020 , 180, 263-277.e20	56.2	46
37	Quantification of transplant-derived circulating cell-free DNA in absence of a donor genotype. <i>PLoS Computational Biology</i> , 2017 , 13, e1005629	5	36
36	High-resolution mapping of cancer cell networks using co-functional interactions. <i>Molecular Systems Biology</i> , 2018 , 14, e8594	12.2	32
35	Mutation Rate Variation is a Primary Determinant of the Distribution of Allele Frequencies in Humans. <i>PLoS Genetics</i> , 2016 , 12, e1006489	6	30
34	msCentipede: Modeling Heterogeneity across Genomic Sites and Replicates Improves Accuracy in the Inference of Transcription Factor Binding. <i>PLoS ONE</i> , 2015 , 10, e0138030	3.7	28
33	Genetics of 38 blood and urine biomarkers in the UK Biobank		25
32	Evidence for Weak Selective Constraint on Human Gene Expression. <i>Genetics</i> , 2019 , 211, 757-772	4	25
31	Reduced signal for polygenic adaptation of height in UK Biobank		24
30	GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background. <i>ELife</i> , 2021 , 10,	8.9	23
29	Reprogramming LCLs to iPSCs Results in Recovery of Donor-Specific Gene Expression Signature. <i>PLoS Genetics</i> , 2015 , 11, e1005216	6	22
28	Frequent nonallelic gene conversion on the human lineage and its effect on the divergence of gene duplicates. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017 , 114, 12779-12784	11.5	22

27	The effect of freeze-thaw cycles on gene expression levels in lymphoblastoid cell lines. <i>PLoS ONE</i> , 2014 , 9, e107166	3.7	18
26	LeafCutter: annotation-free quantification of RNA splicing		18
25	Variable prediction accuracy of polygenic scores within an ancestry group		18
24	Post-translational buffering leads to convergent protein expression levels between primates. <i>Genome Biology</i> , 2018 , 19, 83	18.3	16
23	Response to Comment on "Genetic Structure of Human Populations". <i>Science</i> , 2003 , 300, 1877c-1877	33.3	15
22	Remodeling the Specificity of an Endosomal CORVET Tether Underlies Formation of Regulated Secretory Vesicles in the Ciliate <i>Tetrahymena thermophila</i> . <i>Current Biology</i> , 2018 , 28, 697-710.e13	6.3	14
21	Batch effects and the effective design of single-cell gene expression studies		9
20	Whole Genome Sequencing Identifies a Novel Factor Required for Secretory Granule Maturation in <i>Tetrahymena thermophila</i> . <i>G3: Genes, Genomes, Genetics</i> , 2016 , 6, 2505-16	3.2	8
19	Public Discussion Affects Question Asking at Academic Conferences. <i>American Journal of Human Genetics</i> , 2019 , 105, 189-197	11	7
18	Landscape of stimulation-responsive chromatin across diverse human immune cells		7
17	Shared heritability of human face and brain shape. <i>Nature Genetics</i> , 2021 , 53, 830-839	36.3	7
16	Author response: Variable prediction accuracy of polygenic scores within an ancestry group 2019 ,		6
15	Impact of regulatory variation across human iPSCs and differentiated cells		6
14	Detection of human adaptation during the past 2,000 years		5
13	GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background		5
12	Trans effects on gene expression can drive omnigenic inheritance		5
11	Author response: Determining the genetic basis of anthracycline-cardiotoxicity by molecular response QTL mapping in induced cardiomyocytes 2018 ,		4
10	Systematic discovery and perturbation of regulatory genes in human T cells reveals the architecture of immune networks		4

9	Author response: Reduced signal for polygenic adaptation of height in UK Biobank 2018 ,		3
8	Measurement of selective constraint on human gene expression		3
7	Genetic interactions drive heterogeneity in causal variant effect sizes for gene expression and complex traits		1
6	Adaptive evolution of conserved non-coding elements in mammals. <i>PLoS Genetics</i> , 2005 , preprint, e147	6	1
5	High-resolution mapping of cancer cell networks using co-functional interactions		1
4	Evolutionary persistence of DNA methylation for millions of years after ancient loss of a de novo methyltransferase		
3	Inferring relevant cell types for complex traits using single-cell gene expression		1
2	A Bibliometric History of the Journal GENETICS. <i>Genetics</i> , 2016 , 204, 1337-1342	4	1
1	A natural mutator allele shapes mutation spectrum variation in mice.. <i>Nature</i> , 2022 , 605, 497-502	50.4	1