

# Jonathan K Pritchard

## List of Publications by Year in Descending Order

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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	A natural mutator allele shapes mutation spectrum variation in mice.. <i>Nature</i> , <b>2022</b> , 605, 497-502	50.4	1
133	Shared heritability of human face and brain shape. <i>Nature Genetics</i> , <b>2021</b> , 53, 830-839	36.3	7
132	Genetics of 35 blood and urine biomarkers in the UK Biobank. <i>Nature Genetics</i> , <b>2021</b> , 53, 185-194	36.3	78
131	GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background. <i>ELife</i> , <b>2021</b> , 10,	8.9	23
130	Large-scale cis- and trans-eQTL analyses identify thousands of genetic loci and polygenic scores that regulate blood gene expression. <i>Nature Genetics</i> , <b>2021</b> , 53, 1300-1310	36.3	60
129	Chromatin accessibility dynamics in a model of human forebrain development. <i>Science</i> , <b>2020</b> , 367,	33.3	76
128	Evolutionary Persistence of DNA Methylation for Millions of Years after Ancient Loss of a De Novo Methyltransferase. <i>Cell</i> , <b>2020</b> , 180, 263-277.e20	56.2	46
127	Variable prediction accuracy of polygenic scores within an ancestry group. <i>ELife</i> , <b>2020</b> , 9,	8.9	112
126	Trans Effects on Gene Expression Can Drive Omnigenic Inheritance. <i>Cell</i> , <b>2019</b> , 177, 1022-1034.e6	56.2	183
125	Public Discussion Affects Question Asking at Academic Conferences. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 189-197	11	7
124	Ancient Rome: A genetic crossroads of Europe and the Mediterranean. <i>Science</i> , <b>2019</b> , 366, 708-714	33.3	59
123	Reduced signal for polygenic adaptation of height in UK Biobank. <i>ELife</i> , <b>2019</b> , 8,	8.9	181
122	Author response: Variable prediction accuracy of polygenic scores within an ancestry group <b>2019</b> ,		6
121	Landscape of stimulation-responsive chromatin across diverse human immune cells. <i>Nature Genetics</i> , <b>2019</b> , 51, 1494-1505	36.3	86
120	Evidence for Weak Selective Constraint on Human Gene Expression. <i>Genetics</i> , <b>2019</b> , 211, 757-772	4	25
119	Interpreting polygenic scores, polygenic adaptation, and human phenotypic differences. <i>Evolution, Medicine and Public Health</i> , <b>2019</b> , 2019, 26-34	3	51
118	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> , <b>2018</b> , 28, 697-710.e13	6.3	14

117	Large-Scale Clonal Analysis Resolves Aging of the Mouse Hematopoietic Stem Cell Compartment. <i>Cell Stem Cell</i> , <b>2018</b> , 22, 600-607.e4	18	77
116	Post-translational buffering leads to convergent protein expression levels between primates. <i>Genome Biology</i> , <b>2018</b> , 19, 83	18.3	16
115	Determining the genetic basis of anthracycline-cardiotoxicity by molecular response QTL mapping in induced cardiomyocytes. <i>ELife</i> , <b>2018</b> , 7,	8.9	54
114	Author response: Determining the genetic basis of anthracycline-cardiotoxicity by molecular response QTL mapping in induced cardiomyocytes <b>2018</b> ,		4
113	Author response: Reduced signal for polygenic adaptation of height in UK Biobank <b>2018</b> ,		3
112	Annotation-free quantification of RNA splicing using LeafCutter. <i>Nature Genetics</i> , <b>2018</b> , 50, 151-158	36.3	234
111	Impact of regulatory variation across human iPSCs and differentiated cells. <i>Genome Research</i> , <b>2018</b> , 28, 122-131	9.7	70
110	High-resolution mapping of cancer cell networks using co-functional interactions. <i>Molecular Systems Biology</i> , <b>2018</b> , 14, e8594	12.2	32
109	Functional Genetic Variants Revealed by Massively Parallel Precise Genome Editing. <i>Cell</i> , <b>2018</b> , 175, 544-562.e16	56.2	169
108	Tracing the peopling of the world through genomics. <i>Nature</i> , <b>2017</b> , 541, 302-310	50.4	338
107	An Expanded View of Complex Traits: From Polygenic to Omnigenic. <i>Cell</i> , <b>2017</b> , 169, 1177-1186	56.2	1413
106	Batch effects and the effective design of single-cell gene expression studies. <i>Scientific Reports</i> , <b>2017</b> , 7, 39921	4.9	205
105	Inferring Relevant Cell Types for Complex Traits by Using Single-Cell Gene Expression. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 686-699	11	61
104	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> , <b>2017</b> , 114, 12779-12784	11.5	22
103	Rapid evolution of the human mutation spectrum. <i>ELife</i> , <b>2017</b> , 6,	8.9	82
102	Quantification of transplant-derived circulating cell-free DNA in absence of a donor genotype. <i>PLoS Computational Biology</i> , <b>2017</b> , 13, e1005629	5	36
101	Lineage-specific and single-cell chromatin accessibility charts human hematopoiesis and leukemia evolution. <i>Nature Genetics</i> , <b>2016</b> , 48, 1193-203	36.3	555
100	Abundant contribution of short tandem repeats to gene expression variation in humans. <i>Nature Genetics</i> , <b>2016</b> , 48, 22-9	36.3	184

99	Mutation Rate Variation is a Primary Determinant of the Distribution of Allele Frequencies in Humans. <i>PLoS Genetics</i> , <b>2016</b> , 12, e1006489	6	30
98	Thousands of novel translated open reading frames in humans inferred by ribosome footprint profiling. <i>ELife</i> , <b>2016</b> , 5,	8.9	82
97	Genetic Variation, Not Cell Type of Origin, Underlies the Majority of Identifiable Regulatory Differences in iPSCs. <i>PLoS Genetics</i> , <b>2016</b> , 12, e1005793	6	79
96	Genome-wide association study of behavioral, physiological and gene expression traits in outbred CFW mice. <i>Nature Genetics</i> , <b>2016</b> , 48, 919-26	36.3	72
95	A Bibliometric History of the Journal GENETICS. <i>Genetics</i> , <b>2016</b> , 204, 1337-1342	4	1
94	Whole Genome Sequencing Identifies a Novel Factor Required for Secretory Granule Maturation in <i>Tetrahymena thermophila</i> . <i>G3: Genes, Genomes, Genetics</i> , <b>2016</b> , 6, 2505-16	3.2	8
93	Coregulation of tandem duplicate genes slows evolution of subfunctionalization in mammals. <i>Science</i> , <b>2016</b> , 352, 1009-13	33.3	90
92	RNA splicing is a primary link between genetic variation and disease. <i>Science</i> , <b>2016</b> , 352, 600-4	33.3	357
91	Detection of human adaptation during the past 2000 years. <i>Science</i> , <b>2016</b> , 354, 760-764	33.3	224
90	Genetic variation in MHC proteins is associated with T cell receptor expression biases. <i>Nature Genetics</i> , <b>2016</b> , 48, 995-1002	36.3	79
89	Reprogramming LCLs to iPSCs Results in Recovery of Donor-Specific Gene Expression Signature. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005216	6	22
88	The genetic and mechanistic basis for variation in gene regulation. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1004857	6	96
87	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans. <i>Science</i> , <b>2015</b> , 348, 648-60	33.3	3242
86	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , <b>2015</b> , 348, 666-9	33.3	170
85	Genetic Control of Chromatin States in Humans Involves Local and Distal Chromosomal Interactions. <i>Cell</i> , <b>2015</b> , 162, 1051-65	56.2	240
84	WASP: allele-specific software for robust molecular quantitative trait locus discovery. <i>Nature Methods</i> , <b>2015</b> , 12, 1061-3	21.6	253
83	Genomic variation. Impact of regulatory variation from RNA to protein. <i>Science</i> , <b>2015</b> , 347, 664-7	33.3	276
82	msCentipede: Modeling Heterogeneity across Genomic Sites and Replicates Improves Accuracy in the Inference of Transcription Factor Binding. <i>PLoS ONE</i> , <b>2015</b> , 10, e0138030	3.7	28

81	The deleterious mutation load is insensitive to recent population history. <i>Nature Genetics</i> , <b>2014</b> , 46, 220-46.3	214
80	Admixture facilitates genetic adaptations to high altitude in Tibet. <i>Nature Communications</i> , <b>2014</b> , 5, 3281-7.4	133
79	The effect of freeze-thaw cycles on gene expression levels in lymphoblastoid cell lines. <i>PLoS ONE</i> , <b>2014</b> , 9, e107166	3.7 18
78	The functional consequences of variation in transcription factor binding. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004226	2.6 135
77	Methylation QTLs are associated with coordinated changes in transcription factor binding, histone modifications, and gene expression levels. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004663	6 187
76	fastSTRUCTURE: variational inference of population structure in large SNP data sets. <i>Genetics</i> , <b>2014</b> , 197, 573-89	4 839
75	Identification of genetic variants that affect histone modifications in human cells. <i>Science</i> , <b>2013</b> , 342, 747-9	33.3 331
74	Primate transcript and protein expression levels evolve under compensatory selection pressures. <i>Science</i> , <b>2013</b> , 342, 1100-4	33.3 156
73	Dissecting the regulatory architecture of gene expression QTLs. <i>Genome Biology</i> , <b>2012</b> , 13, R7	18.3 151
72	DNase I sensitivity QTLs are a major determinant of human expression variation. <i>Nature</i> , <b>2012</b> , 482, 390-40.4	479
71	DNA sequence-dependent compartmentalization and silencing of chromatin at the nuclear lamina. <i>Cell</i> , <b>2012</b> , 149, 1474-87	56.2 337
70	Genomics: ENCODE explained. <i>Nature</i> , <b>2012</b> , 489, 52-5	50.4 191
69	The genetic architecture of adaptations to high altitude in Ethiopia. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1003110	6 137
68	A systematic survey of loss-of-function variants in human protein-coding genes. <i>Science</i> , <b>2012</b> , 335, 823-83.3	880
67	The contribution of RNA decay quantitative trait loci to inter-individual variation in steady-state gene expression levels. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1003000	6 80
66	Controls of nucleosome positioning in the human genome. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1003036	6 196
65	Comparative RNA sequencing reveals substantial genetic variation in endangered primates. <i>Genome Research</i> , <b>2012</b> , 22, 602-10	9.7 97
64	DNA methylation patterns associate with genetic and gene expression variation in HapMap cell lines. <i>Genome Biology</i> , <b>2011</b> , 12, R10	18.3 651

63	A genome-wide study of DNA methylation patterns and gene expression levels in multiple human and chimpanzee tissues. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1001316	6	159
62	Accurate inference of transcription factor binding from DNA sequence and chromatin accessibility data. <i>Genome Research</i> , <b>2011</b> , 21, 447-55	9.7	406
61	Adaptations to climate-mediated selective pressures in humans. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1001375	6	207
60	Understanding mechanisms underlying human gene expression variation with RNA sequencing. <i>Nature</i> , <b>2010</b> , 464, 768-72	50.4	993
59	Adaptation - not by sweeps alone. <i>Nature Reviews Genetics</i> , <b>2010</b> , 11, 665-7	30.1	321
58	Noisy splicing drives mRNA isoform diversity in human cells. <i>PLoS Genetics</i> , <b>2010</b> , 6, e1001236	6	203
57	Using environmental correlations to identify loci underlying local adaptation. <i>Genetics</i> , <b>2010</b> , 185, 1411-23	49.7	497
56	The genetics of human adaptation: hard sweeps, soft sweeps, and polygenic adaptation. <i>Current Biology</i> , <b>2010</b> , 20, R208-15	6.3	666
55	Effect of read-mapping biases on detecting allele-specific expression from RNA-sequencing data. <i>Bioinformatics</i> , <b>2009</b> , 25, 3207-12	7.2	390
54	Characterizing natural variation using next-generation sequencing technologies. <i>Trends in Genetics</i> , <b>2009</b> , 25, 463-71	8.5	108
53	Signals of recent positive selection in a worldwide sample of human populations. <i>Genome Research</i> , <b>2009</b> , 19, 826-37	9.7	557
52	Inferring weak population structure with the assistance of sample group information. <i>Molecular Ecology Resources</i> , <b>2009</b> , 9, 1322-32	8.4	2406
51	The role of geography in human adaptation. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000500	6	304
50	Gene expression levels are a target of recent natural selection in the human genome. <i>Molecular Biology and Evolution</i> , <b>2009</b> , 26, 649-58	8.3	73
49	Revealing the architecture of gene regulation: the promise of eQTL studies. <i>Trends in Genetics</i> , <b>2008</b> , 24, 408-15	8.5	366
48	High-resolution mapping of crossovers reveals extensive variation in fine-scale recombination patterns among humans. <i>Science</i> , <b>2008</b> , 319, 1395-8	33.3	287
47	High-resolution mapping of expression-QTLs yields insight into human gene regulation. <i>PLoS Genetics</i> , <b>2008</b> , 4, e1000214	6	456
46	Adaptations to climate in candidate genes for common metabolic disorders. <i>PLoS Genetics</i> , <b>2008</b> , 4, e326	6	204

45	Completing the map of human genetic variation. <i>Nature</i> , <b>2007</b> , 447, 161-5	50.4	153
44	Convergent adaptation of human lactase persistence in Africa and Europe. <i>Nature Genetics</i> , <b>2007</b> , 39, 31-40	36.3	1073
43	Inference of population structure using multilocus genotype data: dominant markers and null alleles. <i>Molecular Ecology Notes</i> , <b>2007</b> , 7, 574-578		2333
42	Overcoming the winner's curse: estimating penetrance parameters from case-control data. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 605-15	11	289
41	A map of recent positive selection in the human genome. <i>PLoS Biology</i> , <b>2006</b> , 4, e72	9.7	1811
40	Sequencing and analysis of Neanderthal genomic DNA. <i>Science</i> , <b>2006</b> , 314, 1113-8	33.3	415
39	Clonal origin and evolution of a transmissible cancer. <i>Cell</i> , <b>2006</b> , 126, 477-87	56.2	299
38	A high-resolution survey of deletion polymorphism in the human genome. <i>Nature Genetics</i> , <b>2006</b> , 38, 75-81	36.3	539
37	A worldwide survey of haplotype variation and linkage disequilibrium in the human genome. <i>Nature Genetics</i> , <b>2006</b> , 38, 1251-60	36.3	406
36	Coalescent-based association mapping and fine mapping of complex trait loci. <i>Genetics</i> , <b>2005</b> , 169, 1071-92		99
35	Confounding from cryptic relatedness in case-control association studies. <i>PLoS Genetics</i> , <b>2005</b> , 1, e32	6	163
34	Clines, clusters, and the effect of study design on the inference of human population structure. <i>PLoS Genetics</i> , <b>2005</b> , 1, e70	6	407
33	Adaptive evolution of conserved non-coding elements in mammals. <i>PLoS Genetics</i> , <b>2005</b> , preprint, e147	6	1
32	Evidence for extensive transmission distortion in the human genome. <i>American Journal of Human Genetics</i> , <b>2004</b> , 74, 62-72	11	94
31	Statistical tests for admixture mapping with case-control and cases-only data. <i>American Journal of Human Genetics</i> , <b>2004</b> , 75, 771-89	11	132
30	Response to Comment on "Genetic Structure of Human Populations". <i>Science</i> , <b>2003</b> , 300, 1877c-1877	33.3	15
29	Haplotype blocks and linkage disequilibrium in the human genome. <i>Nature Reviews Genetics</i> , <b>2003</b> , 4, 587-97	30.1	430
28	Assessing the performance of the haplotype block model of linkage disequilibrium. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 502-15	11	124

27	Informativeness of genetic markers for inference of ancestry. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 1402-22	11	495
26	Traces of human migrations in <i>Helicobacter pylori</i> populations. <i>Science</i> , <b>2003</b> , 299, 1582-5	33.3	764
25	Inference of population structure using multilocus genotype data: linked loci and correlated allele frequencies. <i>Genetics</i> , <b>2003</b> , 164, 1567-87	4	5901
24	Genetic structure of human populations. <i>Science</i> , <b>2002</b> , 298, 2381-5	33.3	1995
23	The allelic architecture of human disease genes: common disease-common variant...or not?. <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 2417-23	5.6	515
22	Case-control studies of association in structured or admixed populations. <i>Theoretical Population Biology</i> , <b>2001</b> , 60, 227-37	1.2	325
21	Are rare variants responsible for susceptibility to complex diseases?. <i>American Journal of Human Genetics</i> , <b>2001</b> , 69, 124-37	11	1171
20	Linkage disequilibrium in humans: models and data. <i>American Journal of Human Genetics</i> , <b>2001</b> , 69, 1-14	11	920
19	Association mapping in structured populations. <i>American Journal of Human Genetics</i> , <b>2000</b> , 67, 170-81	11	1472
18	Inference of population structure using multilocus genotype data. <i>Genetics</i> , <b>2000</b> , 155, 945-59	4	22315
17	Use of unlinked genetic markers to detect population stratification in association studies. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 220-8	11	993
16	Genetic interactions drive heterogeneity in causal variant effect sizes for gene expression and complex traits		1
15	High-resolution mapping of cancer cell networks using co-functional interactions		1
14	LeafCutter: annotation-free quantification of RNA splicing		18
13	Detection of human adaptation during the past 2,000 years		5
12	Batch effects and the effective design of single-cell gene expression studies		9
11	Impact of regulatory variation across human iPSCs and differentiated cells		6
10	Evolutionary persistence of DNA methylation for millions of years after ancient loss of a de novo methyltransferase		



9	GWAS of three molecular traits highlights core genes and pathways alongside a highly polygenic background	5
8	Measurement of selective constraint on human gene expression	3
7	Reduced signal for polygenic adaptation of height in UK Biobank	24
6	Landscape of stimulation-responsive chromatin across diverse human immune cells	7
5	Trans effects on gene expression can drive omnigenic inheritance	5
4	Variable prediction accuracy of polygenic scores within an ancestry group	18
3	Genetics of 38 blood and urine biomarkers in the UK Biobank	25
2	Inferring relevant cell types for complex traits using single-cell gene expression	1
1	Systematic discovery and perturbation of regulatory genes in human T cells reveals the architecture of immune networks	4